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Genetic Testing in Emerging Economies (GenTEE)

Summary Report

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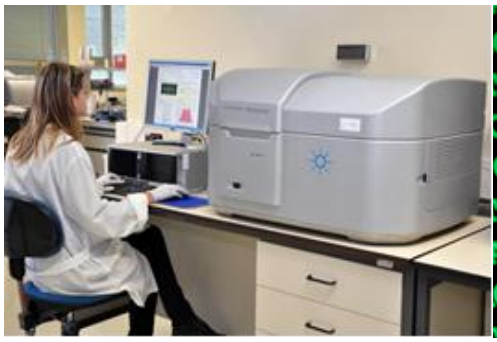

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The GenTEE project is associated with EuroGentest2 (*Genetic Testing in Europe - Network for the further development, harmonization, validation and standardization of services*) work package 8 *"Best Practice Guidelines for Provision of Clinical Genetic Service"*.

Abbreviations

AfrSHG	African Society of Human Genetics
AMB	Associação Médica Brasileira *(Brazilian Medical Association)*
ANVISA	Agência de Vigilância Sanitária *(National Health Surveillance Agency, Brazil)*
ART	Assisted Reproductive Technology
BDRI	Birth Defects Registry of India *(Chennai, India)*
BGI	Beijing Institute of Genomics *(China)*
BRICS	Biotechnology Regional Innovation Centres *(South Africa)*
BSc	Bachelor of Science
CAGS	Centre for Arab Genomic Studies *(Dubai, UAE)*
CAGSE	Concerted Action on Genetics Services in Europe
CAH	Congenital Adrenal Hyperplasia
CANSA	Cancer Association of South Africa
CAP	College of American Pathologists
CAPABILITY	Capacity Building for the Translation of Genetic Knowledge into Practice and Prevention *(Specific Support Action funded by EC, Framework Programme 6)*
CAPES	Coordenação de Aperfeiçoamento de Pessoal de Nível Superior *(Coordination of Improvement of Higher Education Personnel, Brazil)*
CBR	Community-based Rehabilitation
CDC	Centers for Disease Control and Prevention *(Atlanta Georgia, USA)*
CEQA	Cytogenetic European Quality Assessment
CFM	Conselho Federal de Medicina *(Federal Council of Medicine, Brazil)*
CH	Congenital Hypothyroidism
CHED	Commission on Higher Education *(Office of the President of the Philippines, Government of the Philippines)*
CHGC	Chinese National Human Genome Center
CME	Continuing Medical Education
CNPq	Conselho Nacional de Desenvolvimento Científico e Tecnológico *(National Council for Scientific and Technological Development, Brazil)*
CONICET	Consejo Nacional de Investigaciones Científicas y Técnicas *(National Scientific and Technical Research Council, Argentina)*
CSIR	Council of Scientific and Industrial Research (*India*)
DATASUS	Departamento de Informática do Sistema Único de Saúde do Brasil *(Brazilian Ministry of Health database)*
DBT	Department of Biotechnology *(Ministry of Science and Technology, India*)
DM	Doctor of Medicine
DMD	Duchenne Muscular Dystrophy

DNA	Deoxyribonucleic Acid
DoH	Department of Health
DST	Department of Science and *Technology (Ministry of Science and Technology, India*)
EC	European Commission
ECLAMC	Estudio Colaborativo Latino Americano de Malformaciones Congenitas *(Latin American Collaborative Study of Congenital Malformations)*
EMQN	European Molecular Genetics Quality Network
EQA	External Quality Assurance
EU	European Union
EUMEDIS	Euro-MEDiterranean Information Society
EuroGentest	Genetic Testing in Europe - Network for the further development, harmonization, validation and standardization of services
FA	Fanconi anemia
FAP	Fundação de Amparo a Pesquisa *(Foundation for Research Support, Brazil)*
FAPESP	Fundação de Amparo à Pesquisa do Estado de São Paulo *(Foundation for Research Support of São Paulo, Brazil)*
FCMG(SA)	Fellowship of the College of Medical Geneticists *(South Africa)*
FINEP	Financiadora de Estudos e Projetos *(Financier of Studies and Projects, Brazil)*
FISH	Fluorescence In Situ Hybridization
FP	Framework Programme *(European Union)*
G6PD	Glucose-6-Phosphate Dehydrogenase
GBCS	Guangzhou Biobank Cohort Study *(China)*
GENBANK	Genetic Sequence Database *(National Centre for Biotechniology Information, National Institutes of Health, Bethesda, MD, USA)*
Hb AS	Haemoglobin A and haemoglobin S (heterozygous state)
HD	Huntington disease
HDI	Human Development Index
HIV/AIDS	Human immunodeficiency virus/ Acquired Immune Deficiency Syndrome
HPSCA	Health Professions Council of South Africa
HRS	Health Reform System *(Egypt)*
HVP	Human Variome Project
ICGC	International Cancer Genome Consortium
ICMR	Indian Council of Medical Research
IGA	International Genetic Alliance
IGIB	Institute of Genomics and Integrative Biology (*India*)
IHG-NIH	Institute of Human Genetics-National Institutes of Health *(The Philippines)*
IGDD	Indian Genetic Disease Database
INAGEMP	Instituto Nacional de Genética Médica Populacional *(National Institute of Population Medical Genetics, Brazil)*

INCT	Institutos Nacionais de Ciencia e Tecnologia *(National Institutes of Science and Technology, Brazil)*
IPTS	Institute for Prospective Technological Studies *(Seville, Spain)*
ISO	International Organization for Standardization
IVF	In Vitro Fertilisation
KSCDC	Kadoorie Study of Chronic Disease in China *(China Kadoorie Biobank)*
MCI	Medical Council of India
MCT	Ministério da Ciência e Tecnologia *(Ministry of Science and Technology, Brazil)*
MD	Medical Doctor
MIHCL	Law of the People's Republic of China on Maternal and Infant Health Care
MoD	March of Dimes *(White Plains, NY, USA)*
MoH	Ministry of Health
MoHFW	Ministry of Health & Family Welfare *(India)*
MoH&P	Ministry of Health & Population *(Egypt)*
MRC	Medical Research Council *(South Africa)*
MS	Member States *(European Union)*
MSc	Master of Science
MToP	Medical Termination of Pregnancy
NABL	National Accreditation Board for Testing and Calibration Laboratories *(India)*
NDST	National Department of Science and Technology *(South Africa)*
NGO	Non-governmental organization
NGS	Next-Generation Sequencing
NHA	National Health Act *(South Africa)*
NHLS	National Health Laboratory Service *(South Africa)*
NIBMG	National Institute of Biomedical Genomics *(Kalyani, India*)
NIH	National Institutes of Health *(Bethesda, MD, USA)*
NPCDCS	National Program for Prevention and Control of Cancer, Diabetes, CVD and Stroke *(India)*
NRC	National Research Centre *(Cairo, Egypt)*
NRF	National Research Foundation *(Pretoria, South Africa)*
NRHM	National Rural Health Mission (*India*)
NSFC	Natural Science Foundation of China
NSRC	Newborn Screening Reference Center *(The Philippines)*
OECD	Organization for Economic Co-operation and Development
ONSA	Organization for Nucleotide Sequencing and Analysis *(São Paulo, Brazil)*
PAHO	Pan American Health Organization
PBDS	Philippine Birth Defects Surveillance
PCARI	Philippine California Advanced Research Institutes *(The Philippines)*
PGC	Philippine Genome Center *(Manila, The Philippines)*

PGD	Preimplantation Genetic Diagnosis
PGH	Philippine General Hospital *(Manila, The Philippines)*
PHC	Primary Health Care
PhD	Doctor of Philosophy
PKU	Phenylketonuria
PND	Prenatal Diagnosis
PNTN	Programa Nacional de Triagem Neonatal *(National Newborn Screening Programme, Brazil)*
Rede-EIM Brasil	Rede de Erros Inatos do Metabolismo *(Network for Diagnosis in Inborn Errors of Metabolism, Brazil)*
RENAC	Registro Nacional de Anomalías Congénitas *(National Register of Congenital Anomalies, Argentina)*
SABDSS	South Africa Birth Defects Surveillance System
SAMRC	South African Medical Research Council
SANAS	South African National Accreditation System
SASHG	South African Society of Human Genetics
SBG	Sociedade Brasileira de Genética *(Brazilian Society of Genetics)*
SBGM	Sociedade Brasileira de Genética Médica *(Brazilian Society of Medical Genetics)*
SCD	Sickle Cell Disorder
SLMC	St. Luke‘s Medical Center *(Manila, The Philippines)*
SMA	Spinal Muscular Atrophy
SQU	Sultan Qaboos University *(Muscat, Oman)*
SUS	Sistema Unico de Saúde *(Brazilian Unified Health System)*
TANDEM MS	Tandem Mass Spectrometry
TB	Tuberculosis
TRC	The Research Council *(Muscat, Oman)*
TRS	Telegenetics Referral System *(The Philippines)*
UNDP	United Nations Development Programme
UP	University of the Philippines
WHO	World Health Organization
WITS	University of the Witwatersrand *(Johannesburg, South Africa)*

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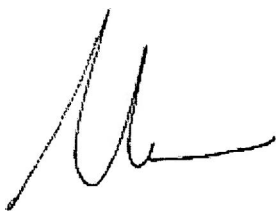
Foreword

It is for me a pleasure to introduce the final report of the GenTEE project, which is the first worldwide effort to systematically survey and assess the current state of medical genetic services in emerging economies.

Due to the epidemiological transition – witnessed by a change in mortality and morbidity patterns – the emerging economies in Asia, Latin America, the Middle East and Africa are facing an increasing proportion of infant morbidity and mortality due to congenital and genetic disorders and an increasing exposure of their adult population to risks for non-communicable chronic diseases such as: heart disease, stroke, cancer and diabetes - diseases that all have subgroups with significant genetic risk components. These changes in risk factors result in an increasing need for genetic services.

The GenTEE international network initiative responds to these challenges by facilitating inter- and intra-country comparison of the current state-of-the-art in the field of genetic services and testing development, with the help of a systematic survey conducted in eight countries selected for their capability and readiness to conduct such a survey including: Argentina, Brazil, China, Egypt, India, Oman, the Philippines and South Africa. Although these countries represent different health care systems and funding schemes, different social structures and cultural backgrounds, they share significant commonalities like the requirement to adjust new demands for essential genetic testing services and for capacity building functions that strategically respond to the needs of those affected by or at risk for congenital/genetic disorders.
Strengthening genetic services is a gradual process that can be facilitated and supported by international networking. This international project stands in the tradition of previous networking projects funded by the European Commission on the collection of comparative data on genetic services, namely the "Concerted Action on Genetics Services in Europe" (CAGSE, funded by FP5), the European Commission Joint Research Centre Institute for Prospective Technological Studies' (IPTS) survey "Towards quality assurance and harmonisation of genetic testing services in the EU", "Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention" (CAPABILITY, funded by FP6) and "Genetic Testing in Europe - Network for the further development, harmonization, validation and standardization of services" (EuroGentest, funded by FP6 and FP7).
One of the key roles of the JRC is to use its neutral position and scientific expertise to contribute through pilot projects and networking, to the foundation for a wide public health policy. The survey has generated important data for key stake-holders and policy makers in the participating countries and initiated collaboration among scientists from EU member states and international non EU cooperation partners. I am glad that the support given by the IHCP so far has contributed significantly to the high visibility and success of GenTEE.

K. Maruszewski
IHCP Director



The Report

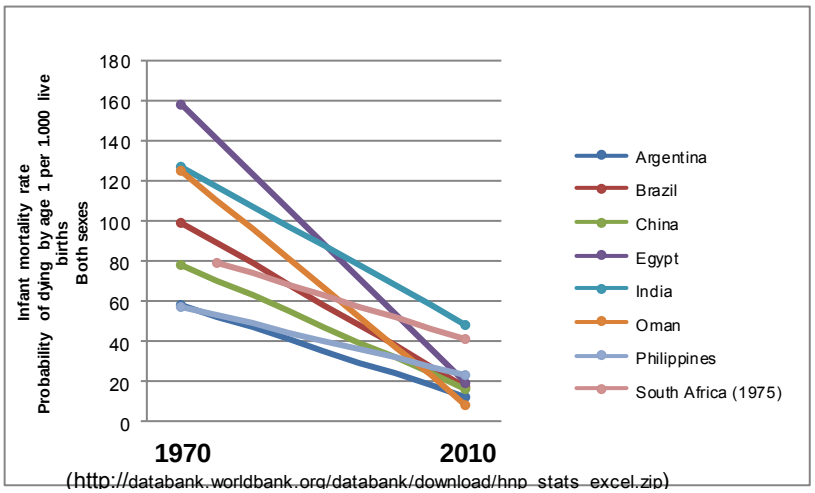
I Background

The GenTEE project is the first project worldwide that systematically reports and assesses the development and current state of medical genetic services in eight emerging economies: ***Argentina***, ***Brazil***, ***China***[1], ***Egypt***, ***India***, ***Oman***, ***Philippines*** and ***South Africa*** (in the following referred to as GenTEE countries). The project is intended to inform policy decisions for the challenges of delivering equitable high quality genetic services and to promote international collaboration for capacity building.

Due to the epidemiological transition in the emerging economies of Asia, Latin America, the Middle East and South Africa, characterized by

(i) a significant reduction of infant mortality from infectious diseases and malnutrition (**Figure 1.1**)

Figure 1.1 Infant mortality rate, probability of dying by age 1 year per 1 000 live births (GenTEE countries, 1970 - 2010)



(http://databank.worldbank.org/databank/download/hnp_stats_excel.zip)

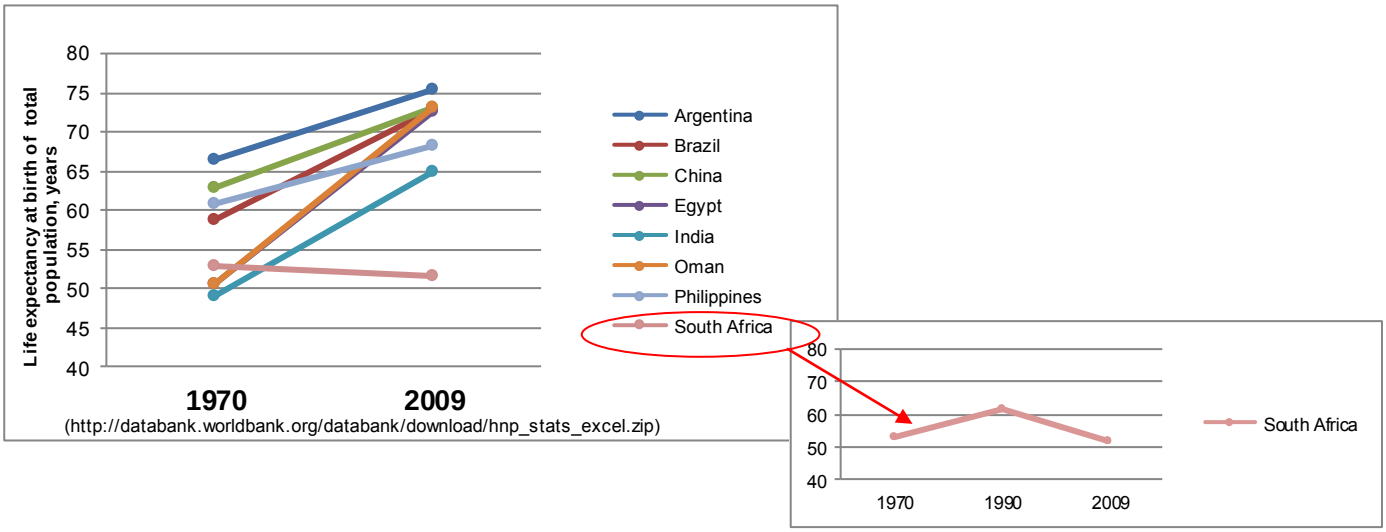
and

(ii) increasing life expectancy[2] (**Figure 1.2**).

[1] Without the Special Administrative Region Hong Kong; The People's Republic of China is referred to throughout the report as China.

[2] South Africa, affected by the HIV/AIDS epidemic, exemplifies that the transition process is not always unidirectional and progress may be halted, reversed or overlapping in different segments of the population.

Figure 1.2 Life expectancy at birth of total population, years (GenTEE countries, 1970 - 2009)[3]



these economies are facing an increasing proportion of infant morbidity and mortality due to congenital and genetic disorders and an increasing exposure of their adult population to risks for non-communicable chronic diseases such as: heart disease, stroke, cancer and diabetes - diseases that all have subgroups with significant genetic risk components.

The changes in risk factors involved in the epidemiological transition result in an increasing need for genetic services to improve both individual patient outcomes and overall population health in these countries.

The challenges the GenTEE emerging economies are facing are manifold:

- develop a service delivery infrastructure, including health workforce training, quality guidelines and procedures leading to equitable and affordable access to high quality genetic/genomic testing services;
- reap the potential benefits that the rapid development of genetic/genomic technologies & knowledge brings and
- ensure the successful translation of genetics/genomics laboratory and academic research into quality assured pathways.

[3] The figures 1.1/1.2: these developments are not linear however this presentation is widely accepted and was adapted from Fineberg H: "A Successful and Sustainable Health System — How to Get There from Here". N Engl J Med 2012;366:1020-7.

The GenTEE Global Network

The GenTEE international network initiative responds to these challenges by facilitating inter- and intra-country comparison on the current state of genetic service and testing development with the help of (i) a systematic international survey conducted in the eight countries[4] selected for their capability and readiness to conduct such a survey and (ii) demonstration projects for capacity building.

Reported here are the concept and the outcome of the international GenTEE survey.

The GenTEE network closely links leading experts in medical genetics from ***Argentina***, ***Brazil***, ***China***, ***Egypt***, ***India***, ***Oman***, ***the Philippines*** and ***South Africa*** (GenTEE countries) with European Union (EU) programmes and institutions that are tasked to develop, harmonize, validate and standardize genetic testing services in the 27 EU Member States (MS) namely: *EuroGentest*[5] and the Institute for Health and Consumer Protection (IHCP), one of the European Commission's (EC) joint research centres.

In addition, the GenTEE network is linked with other international networking activities such as the March of Dimes (MoD) *Global Network for Maternal and Infant Health* and the international parent & patient alliance (*International Genetic Alliance*, IGA).

Figure 1.3 The GenTEE Global Network

[4] The survey was conducted in 2010-2012.
[5] www.eurogentest.org (accessed May 16, 2013)

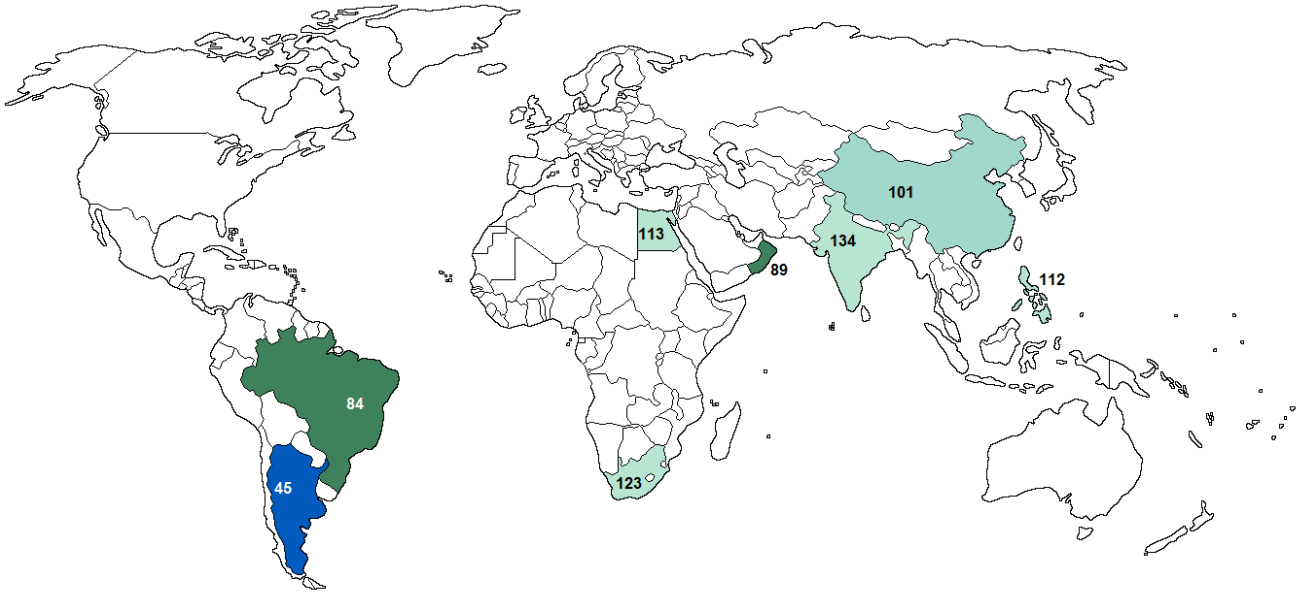
Differences and commonalities

The GenTEE countries are characterised by major differences such as:

- ***geographic size and population size*** ranging from a population of 2.78 million in ***Oman***[6] (2010) to 1.35 billion in ***China***[7] (2010) and a geographical size from 299,764 square kilometres (***the Philippines***, including more than 7000 islands) to 9,596,961 square kilometres (***China***).

- ***levels of development, wealth distribution and income***, representing countries with different HDI ranks such as ***Argentina*** (very high human development), ***Brazil*** and ***Oman*** (high development) and ***China***, ***Egypt***, ***India***, ***the Philippines*** and ***South Africa*** (medium human development) (**Figure 1.4**). The GenTEE country with the highest income is ***Oman*** (high-income country), ***Argentina***, ***Brazil***, ***China*** and ***South Africa*** represent upper-middle income countries and ***Egypt***, ***India*** and ***the Philippines*** represent lower-middle income countries (**Table 1.1**).

Figure 1.4 Human Development Index (HDI): GenTEE countries[8]



Very high human development (rank 1 – 47)

High human development (rank 48 – 94)

[6] World Health Statistics, WHO 2012, p.162. Online available at: http://www.who.int/healthinfo/EN_WHS2012_Full.pdf (accessed January 21, 2013)
[7] World Health Statistics, WHO 2012, p.158. Online available at: http://www.who.int/healthinfo/EN_WHS2012_Full.pdf (accessed January 21, 2013)
[8] Human Development Report 2011: http://hdr.undp.org/en/media/HDR_2011_EN_Complete.pdf (accessed May 16, 2013)

Medium human development (rank 95 – 141)

Table 1.1 Classification of GenTEE countries' economies by the World Bank (2012) [9]

Argentina:	upper-middle-income
Brazil:	upper-middle-income
China:	upper-middle-income
Egypt:	lower-middle-income
India:	lower-middle-income
Oman:	high-income
Philippines:	lower-middle-income
South Africa:	upper-middle-income

GenTEE countries represent different health care systems and funding schemes resulting in different proportions of public vs. private spending. For example, in ***Oman*** [10] public/government expenditure accounts for more than 75% of the total health expenditure and the public sector funds more than 90% of the hospitals, employs most physicians and nurses and is a principal provider of preventive, promotive and rehabilitation services. In other countries such as ***Brazil***, ***China***, ***Egypt***, ***India*** and ***the Philippines*** private expenditure on health is relatively high and consists mostly of direct out-of-pocket based funding (**Table 1.2**).

[9] World Bank, World Bank list of economies, April 2012, source: http://siteresources.worldbank.org/DATASTATISTICS/Resources/CLASS.XLS (accessed May 16, 2013)

[10] For Omani nationals there is universal coverage and health care services are free, for foreigners working in Oman health costs are covered by health insurance through their employers

Table 1.2 Private expenditure on health in GenTEE countries (2009)[11]

country	Private expenditure on health as % of total expenditure on health	Out-of-pocket expenditure as % of private expenditure on health
Argentina	33.6	59.2
Brazil	56.4	57.2
China	47.5	78.9
Egypt	60.5	97.7
India	69.7	86.4
Oman	21.2	63.5
Philippines	64.9	83.6
South Africa	56.2	29.6

Countries represented in the GenTEE survey differ in their cultural backgrounds and include secular countries (***China***, ***South Africa***), countries with strong Catholic traditions (***Argentina***, ***Brazil***, ***the Philippines***), Muslim countries (***Egypt***, ***Oman***) and ***India***, a country where the northern, northeastern and southern parts have distinct ethnic groups, different languages and different religious and cultural traditions. The religious groups vary from the predominant Hindus (80.5%), to Muslims (13.4%), Christians (2.3 %) and Sikhs (1.9%)[12]. Mainland ***China*** has 56 different ethnic groups officially recognized by the government.

GenTEE countries share significant commonalities:

- changing demography and disease patterns;
- increasing congenital disorders/genetic disorder burden;
- need to adjust new demands for essential genetic services;
- need for capacity building functions that strategically respond to the needs of those affected by or at risk for congenital/genetic disorders (and their families), identify unmet needs and support priority choices as needs and given resources determine.

[11] World Health Statistics, WHO 2012, p.134-141. Online available at: http://www.who.int/healthinfo/EN_WHS2012_Full.pdf (accessed January 21, 2013)
[12] http://censusindia.gov.in/Ad_Campaign/drop_in_articles/04-Distribution_by_Religion.pdf (accessed April 9, 2013)

II GenTEE objectives, concept and survey methodology

The GenTEE concept has been developed by the multidisciplinary international GenTEE partnership/consortium representing complementary expertise in medical genetics, genetic epidemiology, genetic testing and service development, quality assessment, genetic education and strategic patient and family-centred advocacy for service improvement.

The GenTEE partners come from European research institutions, international parent and patient organizations and in ***Argentina***, ***Brazil***, ***China***, ***Egypt***, ***India***, ***Oman***, ***the Philippines*** and ***South Africa*** from academia and public health institutions that have been tasked by their national health care systems with policy-planning and service development for the care and prevention of congenital disorders and genetic diseases in their respective countries.

The main objectives of the GenTEE networking project are:

- to document and compare current practices and the state of genetic service provision in the participating GenTEE countries via a standardized survey (GenTEE survey);
- to promote an internationally shared set of basic quality standards for genetic testing and the provision of appropriate genetics/genomics services that will facilitate future joint research, the exchange and transfer of knowledge and new technologies such as high throughput genome analyses via demonstration projects (GenTEE demonstration projects);
- to support joint networking activities by consensus.

The international GenTEE networking project stands in the tradition of previous projects funded by the EC and other international organizations on the collection of comparative data on genetic services development, namely the *"Concerted Action on Genetics Services in Europe"* (CAGSE, funded by FP5) [13], the Institute for Prospective Technological Studies' (IPTS) survey *"Towards quality assurance and harmonisation of genetic testing services in the EU"*[14], the Organisation for Economic Co-operation and Development (OECD) survey *"Quality Assurance and Proficiency Testing for Molecular Genetic Testing"* (2005) [15] and *"Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention"* (CAPABILITY, funded by FP6)[16].

[13] European Journal of Human Genetics 1997; 5 (suppl 2)
[14] http://ipts.jrc.ec.europa.eu/publications/pub.cfm?id=1124 (accessed May 16, 2013)
[15] www.oecd.org/dataoecd/43/6/38839788.pdf (accessed May 16, 2013)
[16] www.http://capabilitynet.eu/ (accessed May 16, 2013)

The GenTEE survey: methods

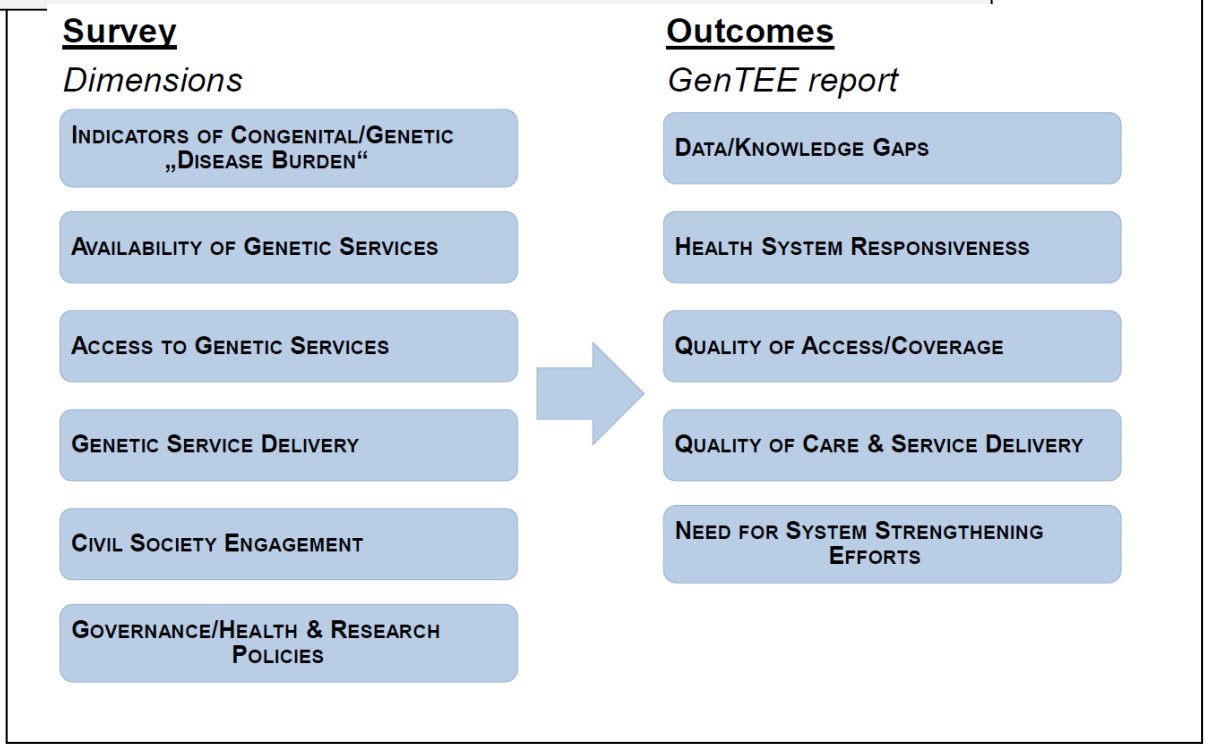
The GenTEE survey

- documents and compares current practices and the state of genetic service provision in participating GenTEE countries;
- identifies current knowledge gaps;
- identifies unmet service needs.

The GenTEE survey is based upon a common method/framework (**Box 2.1**) for data ascertainment, thus allowing examination and comparison of

- service development in the context of a broader view of the existing health care systems;
- given service resources;
- national health policies and genetic testing services development;
- factors related to the state of genetic service delivery such as the demographic, socio-economic and legal factors.

Box 2.1 The GenTEE survey conceptional framework



The GenTEE survey addresses the following key dimensions using a core set of indicators selected by the GenTEE consortium for their relevance and comparability:

- **Demography and health indicators**
  Includes the demographic context in which the health care systems operate, describes variations across countries in population size, birth rates, life expectancy, mortality and other measures of population health status.

- **Health expenditure and financing**
  Compares how much countries spend on health and describes how health services are paid for in the countries (e.g.: public funding, mix between public funding and private health insurance where it exists and out-of-pocket payments by patients).

- **Indicators of congenital/genetic "disorder burden"**
  Documents the available data on birth prevalence of congenital and genetic disorders, and their potential for prevention, known exposure to risk factors that impact birth prevalence and prevalence and specific distribution of congenital and genetic disorders in ethnicities.

- **Availability of genetic services**
  Describes the development (history) of genetic services, reviews the availability of a key set of genetic services for different health purposes and in different settings, including the availability of medical support services such as medical termination of pregnancy (MToP) and counselling services. Addresses trends in genetic testing and provision of service activities and identifies potential drivers for service development.

- **Access to genetic services**
  Describes costs and reimbursement systems, identifies potential barriers to access to genetic services. Potential barriers include: financial barriers (e.g. not being able to afford the costs of a test/service), geographic barriers (e.g. unavailability or paucity of genetic services in a particular area), no timely access (e.g. excessive waiting time), unavailability of particular genetic tests and others barriers.

- **State of genetic services**
  Addresses: available workforce, supply of qualified health personnel, supply and use of technologies, most common type of tests performed and most commonly tested conditions, migration/brain drain, availability of process of "care" recommendations, guidelines and regulatory frameworks.

- **Research priorities in genetics/genomics**
  Addresses current national (government) funding policies and research priorities in genetic/genomics, and research funding by private agencies.

- **Patient organisations and public education in genetics**
  Reviews the availability and structure of parent/patient organisations, including funding, objectives and provision of services.

- **Future outlook for service development in each country**
  Assesses the potential service development taking into account the survey outcome.

The following health care settings for genetic testing services are targeted by the survey:

- Prenatal testing (PND) and pre-implantation genetic diagnosis (PGD);
- Newborn screening;
- Carrier screening;
- Diagnostic testing for congenital and genetic disorders, including testing for common disorders with a major gene subgroup[17];
- Pharmacogenetic testing;
- Genetic susceptibility testing (e.g. for infectious diseases).

Box 2.2 The GenTEE definition of genetic testing

Genetic testing is an analysis of human tissue samples with the aim of detecting or excluding the presence of or risk for particular disorders/conditions with a genetic component.
This includes DNA-based, cytogenetic and biochemical methods.
The purpose of the test should be medical or research related.

Non-medical use or non-genetic use of gene technology are being excluded in the GenTEE survey.

[17] Molecular diagnostic testing for infectious diseases is available in all the countries. This underlines the recognition and acceptance by the GenTEE countries of molecular genetic diagnostic testing in disciplines outside medical genetics.

Data ascertainment

The data collection of the survey in each country is based on:

- published data, including so-called grey-literature[18];
- accessible unpublished reports/data;

  and where deemed necessary:
- expert opinion.

When data are not available for a specific dimension or indicator, this is documented as "unknown/data not available". The survey itself did not set out to generate data when data were not available.

Data for each country are presented as “country reports”[19], each country following the same structure while being able to address individual country characteristics and singular developments. Before a country report was accepted by the GenTEE consortium it had to be submitted to an external expert review for validation.
The country reports were completed (including validation) in 2012.

Single country reports will be published in a forthcoming special issue of the *Journal of Community Genetics* in 2013.

[18] The definition of the term „grey literature“ varies. In the context of the GenTEE survey it is understood as information produced by government, academics, non-government organizations in electronic or print formats that is usually available through specialized channels or the internet and does not enter usual systems of publication distribution and bibliographic or peer review control.
[19] are included in the GenTEE report as appendixes

III Congenital and genetic disorder burden

The survey highlights significant gaps in most GenTEE countries in terms of national epidemiological data availability on congenital and genetic disorders.

Box 3.1 GenTEE definitions of "congenital and genetic disorders"

- *Congenital disorder:* any structural or functional abnormality that is present from birth.[1]
- *Genetic disorder:* a disorder that is typically addressed by medical geneticists through genetic testing, genetic counselling or both, namely constitutive chromosomal abnormalities, single-gene disorders (including the monogenic subgroups of some common disorders) and multifactorial disorders.

[1] The cause may be genetic or due to abnormality in the post conception environment, including teratogens, foetal disruption and constraint.

Availability of national data on congenital and genetic disorder burden

None of the eight GenTEE countries has established ***comprehensive*** population based congenital disorder surveillance systems or registries that document the birth prevalence and population prevalence of congenital and genetic disorders. The lack of national epidemiological data in the GenTEE countries clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders. This in turn impacts severely the capacity to make evidence-informed decisions on planned service development.

Box 3.2 Seven out of the eight GenTEE countries contribute data to international genetic data bases

- ***Argentina*** and ***Brazil*** participate in the ongoing Latin-American Collaborative Study of Congenital Malformations – ECLAMC;
- ***Brazil*** contributes data to the international genetic sequence database (GenBank);
- ***China*** and ***Egypt*** have recently started to report data from hospital-based diagnostic testing laboratories to the Human Variome Project (HVP);
- ***China*** and ***India*** participate in the International Cancer Genome Consortium;
- ***India*** and ***South Africa*** provide data to the International Clearinghouse for Birth Defects. (***South Africa*** provides data on neural tube defects only.);
- ***Oman*** provides data on genetic disorders in ***Oman*** for the Centre for Arab Genomic Studies (CAGS[1]).

[1]Tadmouri GO et al. (2008): Genetic Disorders in Arab World: Oman. Vol 3. Publication of Center for Arab Genomic Studies (CAGS), Dubai, UAE

Most countries have hospital-based registries or surveys that provide figures on the birth prevalence of individual congenital anomalies visible and diagnosable at birth (***Brazil***, ***China***, ***India***, ***Oman***, ***South Africa***). However data may be restricted to individual hospitals, districts, regions or provinces/states (***Brazil, India***), not open for the public (***China***), or discontinued due to lacking funds and priority in the given setting (***South Africa***).

In ***Argentina,*** until recently, the closest estimation of prevalence at birth of congenital defects was that of the 27 congenital malformations registered by ECLAMC, a non-governmental organization (NGO) devoted to develop registries of congenital malformations in South America.[20]

Starting in 2011, the National Ministry of Health (MoH) initiated a *National Registry of Congenital Anomalies* (RENAC), centrally coordinated by the National Medical Genetics Center, an agency of the Ministry. In the period 2009-2011, 182,070 live neonates (28% of the total annual number of births of the country) were examined in 107 hospitals, finding 3,234 neonates with major structural defects (1.78%).[21]

In ***India***, the *Birth Defects Registry of India* (BDRI)[22] based in Chennai gets reports from 309 hospitals from all parts of ***India***. Almost 1 million births have been covered.

[20]. Campaña H, Pawluk MS, López Camelo JS, 2010. ECLAMC Study Group. Birth prevalence of 27 selected congenital anomalies in 7 geographic regions of Argentina. Arch Arg Pediatr 108 (5):409-417. See Table 1 for data on Argentina.
[21] RENAC-Ar (2012). Registro Nacional de Anomaías Congénitas de Argentina. Publication of the Ministry of Health of Argentina.
[22] http://www.fcrf.org.in/bdri_abus.asp (accessed April 15, 2013)

The commonest malformations reported are of the nervous system, comprising 34.8 % of all anomalies.

National health data bases – the problem of underreporting

Some countries such as ***Brazil*** (**Box 3.3**) have instruments which – in theory – could provide accurate data on the birth prevalence of congenital and genetic disorders. In practice however, due to ineffective implementation of the instruments, the reliability of this data is questionable.

Box 3.3 Data on the birth prevalence of congenital anomalies from the Brazilian MoH database (DATASUS)

In 2000, the Brazilian government introduced a new data field in the "Liveborn Declaration", an official document issued by hospitals that would have allowed congenital anomalies present at birth to be registered systematically.

However, analysing data available through DATASUS related to births in Brazil in 2006, only 0.6% of live born infants were registered as having a congenital anomaly, suggesting that congenital anomalies are being underreported. In 2008, the last year available in the database, the figures remained very similar. When comparing data from deaths caused by a congenital anomaly with data from the live born registry, Cunha (2002) demonstrated an underreporting of 60.7% of congenital anomalies on birth certificates. Guerra (2008a, 2008b), who also suggested an underreporting, showed unsatisfactory reliability in the coding of the recorded anomalies by the secretary of health.

Cunha J et al. (2002): Defeitos congênitos em Porto Alegre: uma investigação da qualidade dos dados registrados na Declaração de Nascido Vivo. Revista Brasileira de Epidemiologia (supl.)51.
Guerra FA et al. (2008a): Defeitos congênitos no Município do Rio de Janeiro, Brasil: uma avaliação através do SINASC (2000-2004) Cad. Saúde Pública, Rio de Janeiro, 24(1):140-149.
Guerra FA et al. (2008b): Confiabilidade das informações das declarações de nascido vivo com registro de defeitos congênitos no Município do Rio de Janeiro, Brasil, 2004 Cad. Saúde Pública, Rio de Janeiro, 24(2):438-446

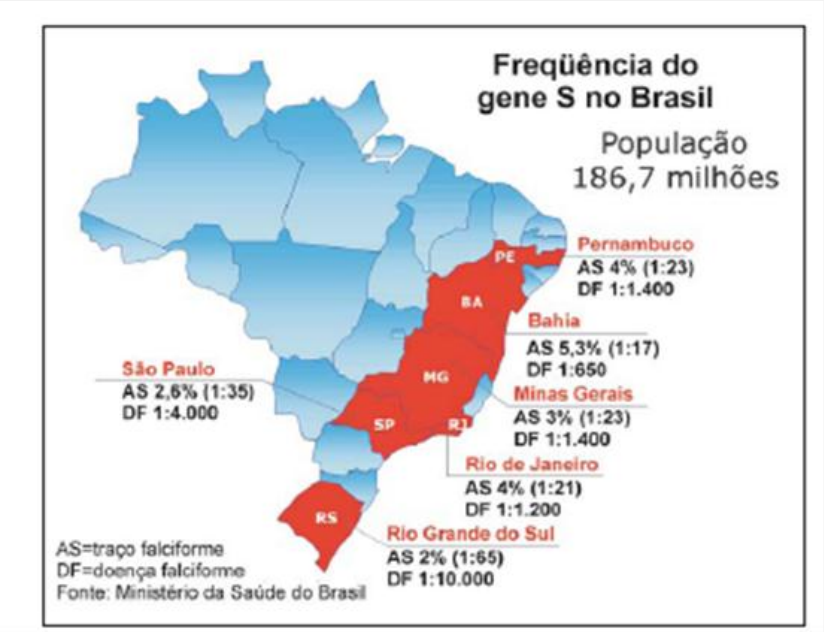
Oman has a *"Central Notification of Birth Defects and Congenital Disorders detectable at Birth"* monitoring system.[23] However, systematic and accurate centralised national data collection is hampered by the marked absence of the correct identification at birth of children with congenital conditions.
Countries with national newborn screening programmes (***Argentina***, ***Brazil***, ***China***, ***Egypt***, ***Oman***, ***the Philippines***) are able to provide data on the birth prevalence of country specific selected disorders including certain autosomal-recessive disorders. However due to restricted coverage of births (coverage may vary among provinces, rural and urban areas), namely in ***Argentina***, ***Brazil***, ***China*** and ***India*** data may be underreporting true national birth prevalence of the disorders reported.

[23] Khandekar R, Jaffer Y. (2010): Incidence and Determinants of Birth Defects and Enzyme Deficiencies among Live Births in Oman: A review of the 2005 National Register. Sultan Qaboos Univ Med J. 10(1):23-30

Box 3.4 Underreporting of cases: Brazil

Limited coverage impacts the availability of national birth prevalence data and may lead to underreporting cases: According to the Brazilian National Newborn Screening Programme (*Programa Nacional de Triagem Neonatal* – PNTN), the sickle cell disease birth prevalence in 2007 was 0.375/1000 live births which would be equivalent to **1140 new cases** (PNTN 2007)[1]. The programme, however, screens for haemoglobin disorders only in selected states. In these states the birth prevalence is 0.493 per 1000 live births which could represent **1496 new cases** in the country if all states were routinely screening for haemoglobin disorders[2]. Since coverage by the national screening programme is not 100% (a safer estimate would be 85% births covered by the programme), nor do these figures include screening covered by private insurance, there could be up to **1760 new yearly cases**.

Figure 3.1 Map of the S gene for sickle cell in Brazil in the states that screen newborns for the disease



AS: sickle cell trait; DF: sickle cell disease. The overall prevalence of AS is estimated as 4% (2-8%) among general population, and 6-10% among afro-descendents. Source: Brazilian Ministry of Health (Cançado Delfini R (2007): Doenças Falciformes. Prática Hospitalar, Ano IX, N° 50, Mar-Abr/2007, available at http://www.scielo.br/pdf/rbhh/v29n3/v29n3a02.pdf (accessed May 16, 2013))

[1] PNTN - Programa Nacional de Triagem Neonatal – relatório 2007 - (National Newborn Screening Program – 2007 report) http://portal.saude.gov.br/portal/arquivos/pdf/INDICADORES_TRIAGEM_NEONATAL.pdf (accessed May 16, 2013)

[2] It must be pointed out that the distribution of the disease is very heterogeneous throughout the country, being more prevalent in the southeast and northeast regions, and less prevalent in the south. Such differences can be explained by the historical composition of the Brazilian population, with migration of African slaves, Europeans and Orientals to specific regions in different colonization periods and economic cycles.

In some countries hospitals keep data on all their congenital and genetic disorder diagnosed patients (***China***, ***Egypt***, ***India***, ***Oman***), while others have funded single population based studies to obtain data on the birth prevalence and prevalence of specific disorders. In particular ***Oman***, where communicable diseases are successfully controlled, has produced a wealth of data on autosomal recessive disorders which cumulatively, are "common" in ***Oman***. This reflects the situation in a traditional Muslim community with a higher rate of consanguinity and where prevention measures for genetic disorders are still in a preparatory phase. Autosomal recessive disorders are thus major contributors to childhood morbidity and mortality constituting a specific disorder burden for ***Oman***.

In ***India*** neonatological services are well established. The *National Neonatoloy Forum*[24], a country wide organization of neonatologists in ***India***, maintains a perinatal database which records causes of neonatal mortality. Visible malformations are documented. During 2002-2003 data were recorded from 151,436 deliveries. 145,623 were live births and 5,813 were stillbirths. Malformations accounted for 9.2 % of all neonatal deaths, and 7.9 % of stillbirths.

Estimated birth prevalence rates for congenital disorders

The GenTEE national reports provide an overview on the current state of the availability of data on congenital and genetic disorders and importantly document the current data knowledge gaps. Underreporting of congenital and genetic disorders is common in all the countries due to a lack of skilled professionals and limited laboratory services to confirm diagnoses. This results in an underestimation of the birth prevalence, prevalence and burden of disease of congenital and genetic disorders.

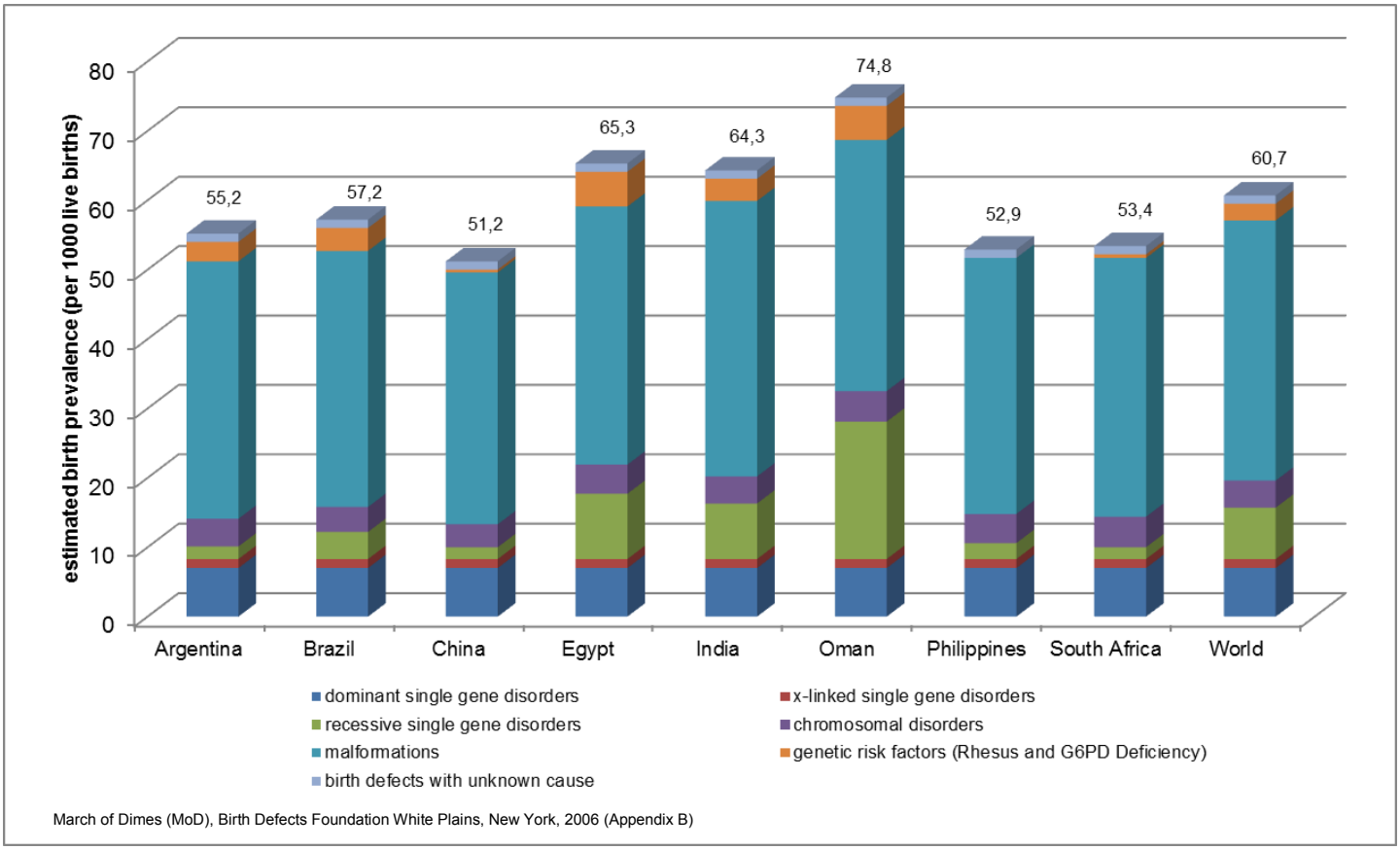
The GenTEE partner MoD in collaboration with South African GenTEE partner A. Christianson documented, in the 2006 MoD *Global Report on Birth Defects*[25], global attention to this data deficit. In order to close the knowledge gap they provided modeled estimates of birth prevalence of congenital disorders, of genetic or partly genetic origin (multifactorial congenital disorders), for 193 countries. These data demonstrate the global impact of congenital disorders.

This is detailed pictorially for the eight GenTEE nations in **Figure 3.2** below. The major difference between nations that contributes to higher birth prevalence of congenital disorders is the birth prevalence of infants with autosomal recessive disorders, particularly conditions with intellectual disability, sickle cell disorder (SCD) and thalassaemia. High levels of consanguinity exacerbate the situation as

[24] www.nnfi.org (accessed May 16, 2013)
[25] http://www.marchofdimes.com/downloads/birth_defects_report-pf.pdf (accessed April 9, 2013)

evidenced in ***Egypt***, ***India*** and ***Oman***. The birth of infants who develop complications secondary to genetic risk factors, essentially Rhesus incompatibility between the parents but especially the presence of Glucose-6-phosphate dehydrogenase (G6PD) deficiency are another factor contributing to high birth prevalence (In ***Oman*** 28% of males and 12% of females have G6PD deficiency but only very small proportion (0.1%) of newborns have complications. There are more than 80 types of G6PD deficiency, most are A+ and B+ types which are innocent. Only a few Mediterranean types lead to haemolysis and neonatal jaundice in newborns).

Figure 3.2 Minimum global estimates of birth prevalence of congenital disorders of genetic or partly genetic origin



March of Dimes (MoD), Birth Defects Foundation White Plains, New York, 2006 (Appendix B)

Figure 3.2 demonstrates that ***Egypt, India and Oman*** are particularly affected by a high birth prevalence of congenital and genetic disorders. It is estimated that ***India*** probably has the largest number of affected infants in the world. Below an estimate of the birth prevalence of selected congenital and genetic disorders has been made for the GenTEE survey based upon hospital-based data (**Box 3.5**).

Box 3.5 Burden of congenital and genetic disorders, national estimate for the birth prevalence of selected disorders for India

Below an estimate of the birth prevalence of congenital and genetic disorders has been made based on published reports.

Disorder	Estimated prevalence/ 1,000 live births	Number of births/year, estimates 2010
Cong. malformations	20	545,400
Down syndrome	1.25	34,000
Metabolic disorders	0.83	22,700
ß-thalassaemia + SCD	0.37	10,100
CH	0.4	11,000
DMD	0.2	5,450
SMA	0.1	2,700

Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.

Impact of congenital and genetic disorders: challenges for national health services in GenTEE countries

India probably has the largest number of carriers for haemoglobinopathies in the world. The need for a national haemoglobinopathies care and prevention programme has been on the agenda of the public debate in ***India*** for decades but yet needs to be addressed by the government of ***India***. The implementation of such a programme on a nationwide scale is met by daunting challenges as outlined in **Box 3.6** below.

Box 3.6 ß-thalassaemia in India

Epidemiology:

The prevalence of carriers varies from 1 - 17% in different ethnic groups with an overall estimate rate of carriers of 3 - 4% (35 - 45 million carriers).
~10,000 - 20,000 affected babies are estimated to be born annually.
~ 100,000 affected children are estimated to be in the country. National survival data are not available.
More than 65 mutations have been characterized in the population, 7 of these mutations account for ~85 - 90% of all mutations identified.[1]

Treatment & care:

The majority of affected children have severe symptoms and receive sub-optimal treatment and clinical management and most of them are not receiving regular blood transfusions and iron chelation. This is mainly due to resource poor clinical settings, limited availability of facilities in the public domain and because the majority of families' lack of financial resources.[2,3]
Bone marrow transplantation is available at few centres but is unaffordable by most families.

Prevention, barriers and constraints:

Although haemoglobinopathies pose a major health problem and health professionals and parent/patient organizations have lobbied for decades for a *"National Haemoglobinopathies Control Programme"*, a national programme is still lacking. India is faced with the challenge how to reach out to a multi-ethnic population of more than 1.2 billion people where the vast majority (> 70%) live in rural areas, only 65% (of the females) and 82 % (of the males) are literate[4]. India is home to 22 scheduled and several hundred other spoken languages[5] and there are 4,693 endogamous communities including 427 tribal groups
The lack of economic and financial resources and insufficient health care infrastructure in the public domain is coupled with a lack of awareness about thalassemia in the public and among health professionals. Reluctance to take up screening offers due to fear of stigmatization has been reported from high risk tribal populations.[6,7]

Introducing services in a gradual way:

In the absence of a national haemoglobinopathy care and prevention programme the ICMR has taken the lead and funded extensive regional multi-centre studies to determine the prevalence of ß-thalassaemia and sickle cell anaemia and to assess the feasibility to establish centres for screening and counselling in medical centres and other institutions where thalassemia facilities are lacking. It is expected that pilot programmes will be implemented in Delhi, Chandigarh and Punjab and that eventually the programme will be integrated into the public health care system in all states.

[1] Colah, R: Control, Strategies for Hemoglobinopathies in India. 1st Pan-Asian Conference on Haemoglobinopathies, country reports. Thalassemia Report 2012; 2 (s1), p.1-2.
[2] Mohanty D et al.: Prevalence of ß-thalassemia and other haemoglobinopathies in six cities in India: a multicentre study. Journal of Community Genetics, October 2012, 4:33-42.
[3] Madan et al.: Frequency of ß-thalassaemia trait and other hemoglobinopathies in northern and western India. Indian Journal of Human Genetics, 2010 Jan-Apr; 16(1): 16-25.
[4] Population Census India 2011. Online available at http://www.census2011.co.in/literacy.php (accessed May 2, 2013)
[5] Verma IC, Saxena R, Kohli S: Past, present & future scenario of thalassaemic care & control in India. Indian Journal of Medical Research 134, October 2011, pp. 507-521.
[6] Mohanty D, Das K: Genetic counselling in tribals in India. Indian Journal of Medical Research 134, October 2011; pp. 561-571.

Oman has enough empiric national data of birth prevalence for congenital and genetic disorders to assist it in planning future medical genetic health services. Using this and modeled birth prevalence data the country has developed projections of annual costs of treating common disorders such as haemoglobin disorders in the absence of primary prevention (**Box 3.7**).

Box 3.7 Projected birth prevalence of congenital and genetic disorders and its impact in Oman

In just three decades Oman has successfully controlled and eradicated major communicable diseases. Whereas in the past the problem of congenital and genetic disorders was hidden in the high mortality rate because most affected infant died before being diagnosed, today more and more infants are diagnosed and provided with the best possible care. In the absence of primary prevention, an increasing number will move into adolescence and adult life in the next years, with important service implications.

Figure 3.3 Oman: projected births of infants with congenital and genetic disorders, 2001 fertility rates

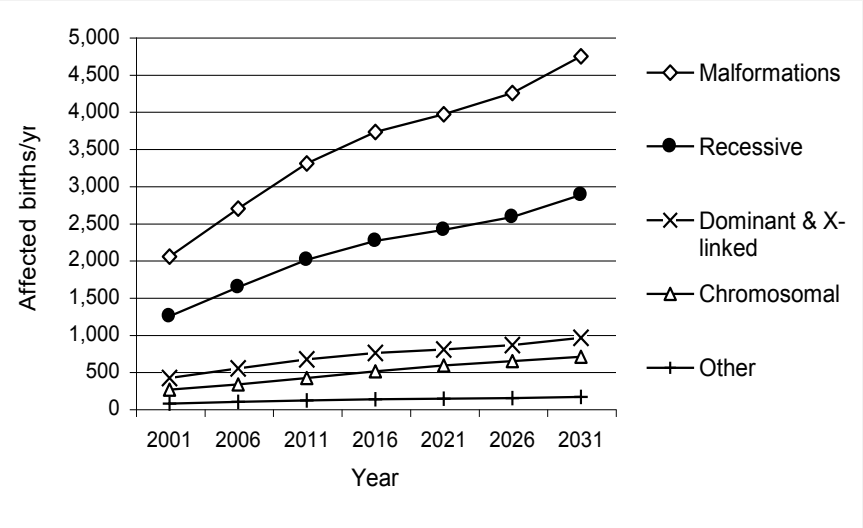


Figure 3.4 Oman: projected annual costs of treating haemoglobin disorders in Oman

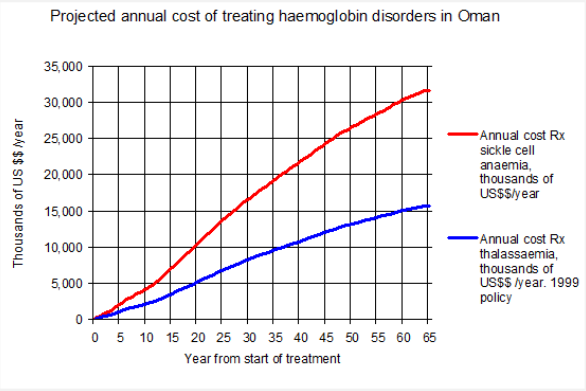
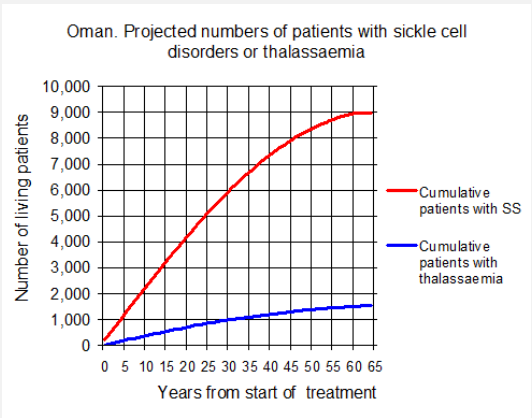


Figure 3.5 Oman: projected numbers of patients with sickle cell disorders or thalassemia



Figures modeled by B. Modell based on data provided by Anna Rajab, Genetic Unit, Directorate General of Health Affairs, Ministry of Health, Muscat, Sultanate of Oman

Congenital and genetic disease burden: infant deaths and under-5 mortality

The data presented in **Box 3.7** below highlight the limited availability and paucity of national data on infant deaths due to congenital disorders in most GenTEE countries. In addition, they demonstrate differences in the national ascertainment of data and the structural weakness in data ascertainment on the impact of congenital/genetic disorders in general. This makes the comparison of data difficult. The table below shows that ***India*** and ***the Philippines*** present the percentage of infant deaths due to congenital/genetic disorders as percentage of neonatal deaths. Whereby ***India*** is probably underreporting deaths considering the World Health Organization (WHO) estimates of 8% of neonatal deaths due to congenital disorders (**Box 3.8**). ***China***, ***Egypt*** and ***South Africa*** cannot provide national data and ***Oman***[26] can only provide data on hospital-based perinatal deaths which include stillbirths.

Box 3.8 Best available national data on infant deaths due to congenital/genetic disorders

	%	year
Argentina	24.7	2011
Brazil	19.3	2010 (congenital anomalies only)
China	unknown/data not available	
Egypt	unknown/data not available	
India	estimate 5.8	2007 (neonatal deaths)
Oman	39	2008 (hospital-based perinatal deaths)
Philippines	11.3	2005 (neonatal deaths)
South Africa	unknown/data not available	

References

Argentina — Report of the Dirección de Estadística e Información de Salud of the Ministry of Health of Argentina (2011), p. 35, available at www.deis.gov.ar (accessed May 16, 2013)

Brazil — Brasil (2013), Ministério da Saúde, Departamento de Informação e Informática do SUS – DATASUS, Sistema de Informações de Saúde. Mortalidade infantil (menores de um ano) http://tabnet.datasus.gov.br/cgi/tabcgi.exe?sim/cnv/inf10uf.def (accessed January 29, 2013)

India — Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.

Oman — 2009 Annual Health Report of Ministry of Health prepared by Department of Health Information & Statistics.

Philippines — Department of Health Statistics 2006. available at http://www.doh.gov.ph/kp/statistics/infant_deaths.html (accessed May 16, 2013)

[26] In Oman stillbirth rate in Ministry of Health institutions where 95% of national deliveries take place is 7.9 per 100 births

The higher percentages of infant deaths attributed to congenital/genetic disorders in ***Argentina*** and ***Brazil*** are also attributable to the countries having advanced well through epidemiological transition, thus having adequate neonatal care with reduced numbers of deaths from prematurity and infection, with resulting exposure of the deaths due to congenital/genetic disorders. This would also be an issue in ***Oman***, were the deaths due to congenital/genetic disorders differentiated.

The countries with the highest percentage of deaths due to congenital disorders namely ***Argentina***, ***Brazil***, ***Egypt*** and ***Oman*** are the countries that have advanced the most through epidemiological transition. This is indicated in **Figures 1.1 & 1.2** which indicates in 2010 their infant mortality rates were ≤20 per 1,000 live births, and the life expectancy was >70 years. The percentage of deaths due to congenital disorders would be expected to be high in these countries. The number of deaths due to infectious diseases and malnutrition would have dropped significantly in the preceding decades whilst deaths due to 'congenital anomalies' would have remained constant, thus increasing the percentage of deaths due to congenital disorders.

The best available international data (country estimates) on under-5 mortality have been published by WHO.

Figure 3.6 WHO data (country estimates) on congenital "anomalies"[27] as cause of deaths in children under-5 in GenTEE countries (2008, 2010)[28]

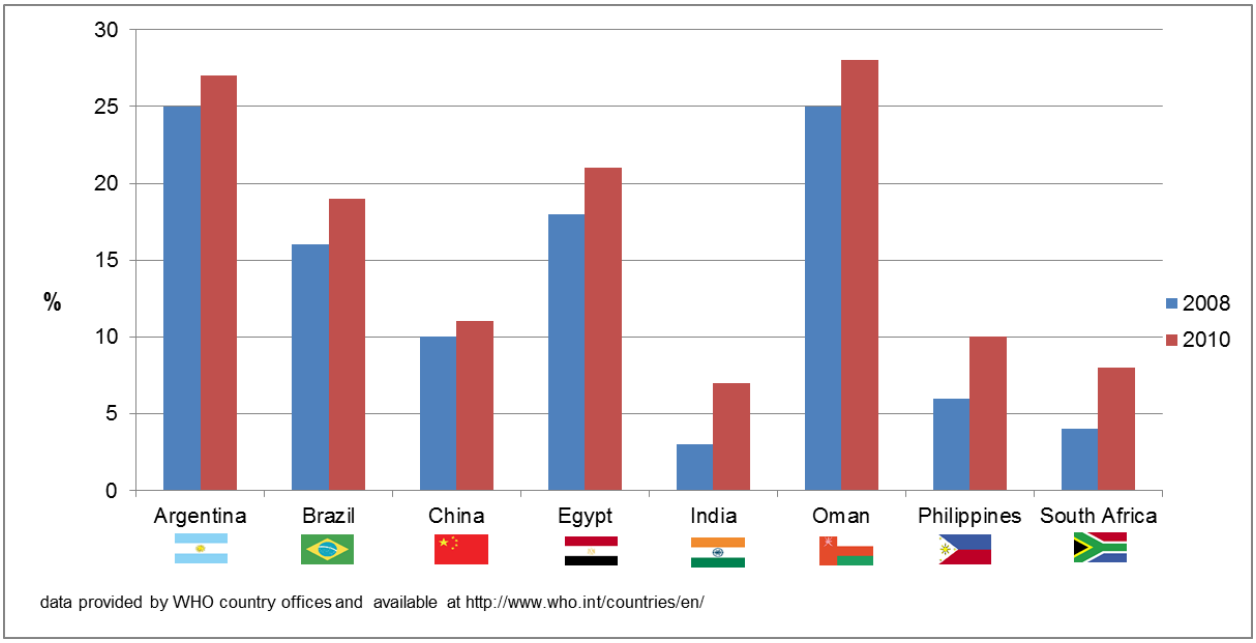


Figure 3.6 confirms the increase of the percentage of deaths due to congenital "anomalies" (disorders) in children under 5 in all GenTEE countries between 2008 and 2010. The data suggest in all countries a temporal trend of an increasing percentage of childhood deaths due to congenital "anomalies", indicating they are currently all experiencing positive epidemiological transition. The highest percentages are reported for ***Oman*** (2008: 25%, 2010: 28%) and ***Argentina*** (2008: 25%, 2010: 27%), followed by ***Egypt*** (2008: 18%, 2010: 21%) and ***Brazil*** (2008: 16%, 2010: 19%).
Oman and ***Argentina*** are nearing the percentages reported in Western countries on congenital "anomalies" as cause of deaths in children under-5.[29]

In contrast, the relatively low percentage of deaths due to congenital disorders in ***India*** (2008: 3%, 2010: 7%), ***the Philippines*** (2008: 6%, 2010: 10%) and ***South Africa*** (2008: 4%, 2010: 8%) could have two reasons. Firstly, and this is particularly true for ***India*** and ***South Africa***, while congenital and genetic disorders have become

[27] The term „anomalies" is employed by WHO.
[28] The WHO data are country level estimates of child deaths under 5 years of age by cause for Member States for the years 2008 and 2010. They represent the best estimates based on the evidence available to it up to the end of 2009 rather than the official estimates oft he Member States. Underlying causes of deaths are defined according to the International Classification of Diseases (ICD). For China a model was developed to assign the total number of child deaths and the main causes of child deaths based upon a total of 206 community based longitudinal studies. For India data are based on a nationally representative sample of over 110,000 deaths in 2001-2003. (WHO: Child mortality by cause, available at http://apps.who.int/gho/data/view.main.gbdc-CH15?lang=en, accessed May 16, 2013)
[29] Western countries with low under-5 deaths (3%) such as Finland, Japan and Norway reported in 2010 36%, 40% and 33% of under-5 deaths due to congenital „anomalies" respectively.

a major disease burden, the number of under-5 year deaths due to infectious diseases and malnutrition remain high.[30] Secondly, there is probably underreporting of deaths due to 'congenital anomalies' due to poor universal clinical diagnostic services and inadequate surveillance and reporting systems. Nevertheless, also in these countries the percentages of deaths due to congenital anomalies are rising significantly albeit from a relatively low percentage.

Congenital and genetic disorders and their impact on paediatric hospital admissions

Data on hospital admissions due to congenital/genetic disorders are scarce in GenTEE countries. Only ***India*** and ***Brazil*** have some hospital-based data. The data ascertained by the Sir Ganga Ram Hospital[31], New Delhi, in 2006, show an admission rate of 8% to its paediatric unit **(Table 3.1)**. Representing the following disorders: 67.3% congenital malformations, 12.2% inborn errors of metabolism, 10.2% single gene disorders and 10.2% chromosomal disorders.

Table 3.1 Genetic disorders/congenital malformations/inborn errors of metabolism: Neonatal Intensive Care Unit, Sir Ganga Ram Hospital, New Delhi, 2006

Disorder	Total no.	%
Total	1222	
All genetic disorders	98	8.0
Malformations	66	67.3
Inborn errors of metabolism	12	12.2
Single gene disorders	10	10.2
(G6PD 7, CAH 1, Albinism 1, Muscle disease 1)		
Chromosomal disorders	10	10.2

Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.

[30] The highest percentage of under-5 deaths in South Africa in 2010 was 28% due to HIV/AIDS and 24% in India due to Pneumonia.

[31] Sir Ganga Ram Hospital is a 650-bed multi-specialty private hospital in New Delhi, India. It provides comprehensive medical services to patients from Delhi and neighbouring states. The hospital has a strong charitable character. Funds generated from the hospital services are partially utilised for providing free health care to the poor and needy patients. All development activities of the hospital are financed from internal resources, with no financial assistance provided by the government or other external agencies.
20% of the beds are available free for admission (including boarding, lodging, investigations, medicine and operative procedures), in addition it runs OPDs where patients are seen free of charge.

For ***Brazil***, admission rates for ages 0-19 years for all hospitals are 2.46% for congenital anomalies. However, in tertiary care hospitals, where collection of data is more specific, of the three main diagnoses coded upon admission congenital and genetic disorders accounted for more than one third of total paediatric admissions.[32,33]

For ***Oman***, only an expert estimate of 50% could be provided for admission rates due to congenital and genetic disorders to the Royal Hospital, Muscat – a tertiary care hospital.

[32] Horovitz DDG. Atenção aos defeitos congênitos no Brasil: propostas para estruturação e integração da abordagem no sistema de saúde [Tese de Doutorado]. Rio de Janeiro: Instituto de Medicina Social, Universidade do Estado do Rio de Janeiro; 2003.
[33] Horovitz DDG, Llerena Jr JC, Mattos RA. Atenção aos defeitos congênitos no Brasil: panorama atual. Cadernos de Saúde Pública 2005; 21(4):1055-1064.

Birth prevalence of selected country specific "common" recessive single gene disorders and distribution of single gene disorders in ethnicities/geographical clusters

GenTEE countries do not have nationwide studies that provide data that would allow the estimate of selected country specific common recessive single gene disorders.

However, in countries such as ***Argentina***, ***Brazil*** and ***the Philippines*** where national newborn screening programmes detect specific autosomal recessive disorders, data about the birth prevalence are available although data may only account for specific regions as coverage varies (**Box 3.4**, Underreporting of cases; **Box 3.5**).

In ***Argentina*** there are no country specific common recessive single gene disorders. Newborn screening detects three autosomal recessive conditions:
Phenylketonuria (PKU) 1/30,000, cystic fibrosis (CF) 1/9,000 and congenital adrenal hyperplasia (CAH) 1/15,000.[34] Geographical clusters of single-gene disorders, usually due to founder effects, have been described but are rare.[35] Two of the most conspicuous are Sandhoff disease in Cordoba province[36] and oculocutaneous albinism in La Roja province.[37] ß-thalassaemia is a common recessive disease as Italian ancestry is reported by close to 50% of the population however, there are no birth prevalence data, and no clusters have been described.[38]

In ***Brazil***, newborn screening detects PKU, sickle cell (Hb AS) and CF. The prevalence of sickle cell trait (Hb AS) is 4% (2-8%) among the general population and 6-10% among Afro-descendants. Estimated population prevalence would be around 7,200,000 Hb AS individuals, 25,000-30,000 with sickle cell disease and 3,500 new cases diagnosed each year.[39] CF is officially screened for in only three Brazilian states, if the birth prevalence considers exclusively births in such areas, it can be estimated as 1/10,000. The demographic composition in these states includes descendants from European immigration (Italy and Germany) and does not reflect ***Brazil*** as a whole. Regarding PKU, estimated birth prevalence by the screening programme is 1/23,000 births, although birth prevalence also varies among regions.[18]

[34] Penchaszadeh, V (2012) GenTEE Report on Argentina.
[35] Castilla EE, Sod R (1990). The surveillance of birth defects in South America. II. The search for geographic clusters: Endemics. Adv Mutag Res 2:211-230.
[36] Dodelson de Kremer R, Depetris de Boldini C, Paschini de Capra A, Pons de Veritier P, Goldenhersch H, Corbella L, Sembaj A, Martín S, Kremer I, Mass L, et al. (1987) Estimation of heterozygote frequency of Sandhoff disease in a high-risk Argentinian population. Predictive assignment of the genotype through statistical analysis. Medicina (Buenos Aires) 47(5):455-63.
[37] Castilla EE, Adams J (1996). Genealogical information and the structure of rural Latin-American populations: reality and fantasy. Hum Hered. 46(5):241-55, 1996.
[38] Feliu-Torres A, Bonduel M, Schiuccati G, del Pozo A et al (2002). Beta talasemia en la Argentina. Medicina 62: 124-134.
[39] PNTN - Programa Nacional de Triagem Neonatal – relatório 2007 - (National Newborn Screening Program – 2007 report) http://portal.saude.gov.br/portal/arquivos/pdf/INDICADORES_TRIAGEM_NEONATAL.pdf (accessed May 16, 2013)

Presently there are some clusters under investigation, as for instance a very high concentration of mucopolysaccharidosis type VI in an isolated region of the state of Bahia.

In ***the Philippines***, the *Newborn Screening Reference Center* (NSRC) documents the following prevalence at birth rates (from 1996 to 2011/ 3,106,938 newborns screened): CAH 1/10,604, Congenital hypothyroidism (CH) 1/3,004 and G6PD deficiency 1/50. [40] Data on the distribution of single gene disorders in ethnicities/geographical clusters are not available for ***the Philippines***.

China, ***Egypt***, ***India***, ***Oman*** and ***South Africa*** have data based on single or multicentre studies or hospital-based data.

In ***China***, data are available from studies that have been conducted among local hospitals. These studies show that the provinces of Guangdong, Guangxi and Hainan are probably the most affected by thalassaemia with an estimated carrier frequency of more than 20% in these areas. The birth prevalence of G6PD deficiency varies in regions and ethnic groups. Again, the birth prevalence seems to be high in southern ***China***, mainly in the provinces of Hainan, Guangdong and Sichuan. The data also show that the prevalence of G6PD deficiency varies among five ethnic groups (Han, Zhuang, Yao, Dai and Jinou). Higher rates were found among the Zhuang, Yao, Dai and Jinou minorities as compared to the Han majority.

In ***Egypt***, studies carried out by the Ministry of Health & Population (MoH&P) Children with Special Needs Department in three different governorates to assess the frequency of ß-thalassaemia carriers among secondary school students showed that frequency rates varied among the governorates. The highest frequency was found in Cairo (1.95%), the lowest in Qaliobia (0.87%).

In ***India***, regional studies conducted under the aegis of Indian Council of Medical Research (ICMR) showed that the carrier frequency of the gene for ß-thalassaemia varies in different regions and in different communities. On the average it is estimated that 3-4 % of people in ***India*** are carriers of the ß-thalassaemia gene (i.e. one in 30/45 people in ***India*** are carriers of ß-thalassaemia). This will calculate to 35-45 million carriers. The frequency is higher in north and western ***India***, and less in south ***India***. In high risk states the carrier frequency is about 5 %. This will calculate to a birth prevalence of ß-thalassaemia major to be 0.63 per 1,000 live births (1/1,600).

The prevalence of Hb E varies among the different caste/ethnic groups in ***India*** and the carrier frequency may be as high as 30-40% in tribal communities.

[40] Padilla CD, de la Paz, EM: Genetic services and testing in the Philippines. Journal of Community Genetics. Online first. July 22, 2012. Online available at http://link.springer.com/content/pdf/10.1007%2Fs12687-012-0102-4.pdf (accessed May 2, 2013)

According to a recent report presented by R. Colah: *"sickle cell anemia is a major problem in central India and the tribal belts in the west, east and south especially in the states of Madhya Pradesh, Chattisgarh, South Gujarat, Maharashtra and Orissa. Although it is mainly seen among tribal population groups it is also present in some non-tribal communities. The prevalence of sickle cell carriers is as high as 30-40% in some of these population groups. Estimates indicate that more than 5000 babies with sickle cell anemia would be born each year."*[41]

The information collected from the various genetic centers shows that common single gene disorders, in addition to thalassaemia, are spinal muscular atrophy (SMA), sensorineural deafness, albinism, CAH, Wilson disease, mucopolysaccharidosis, galactosemia and CF.[42,43,44]

For ***Oman***, hospital data including 420,000 live births are available.[45] These data show that autosomal recessive disorders are common in ***Oman***. Although most of these autosomal recessive disorders are rare, they add up to a large number when these rare disorders are totaled together.

In ***South Africa***, studies showed that inherited autosomal recessive conditions of unusual prevalence were found among Blacks (SMA, oculocutaneous albinism, Fanconi anemia (FA)), Black immigrants (sickle cell anaemia), Ashkenazi Jews (Gaucher disease, Tay Sachs disease), White (CF), Greek and Indian immigrants (thalassaemia) and Afrikaners (Polycystic kidney disease, FA).
Clustered geographic distributions of single gene disorders were reported by six countries (**Box 3.9**). Ethnic clusters were reported by ***China***, ***India*** and ***South Africa***.

[41] Colah, R: Control, Strategies for Hemoglobinopathies in India. 1st Pan-Asian Conference on Haemoglobinopathies, country reports. Thalassemia Report 2012; 2 (s1), p.1-2.
[42] Verma IC, Saxena R, Lall M, Sharma RI. Genetic counseling and prenatal diagnosis in India- experience at Sir Ganga Ram Hospital. Indian J Pediatr 2003; 70: 293-297.
[43] Puri RD, verma IC. Genetic services in India. : A model for developing countries. In "Genomics and Health in the Developing World.' Edited by Kumar D. Oxford University Press, Oxford. 2012. pp 927-35.
[44] Verma IC, Kumar D. Epidemiology of genetic diseases in the Indian subcontinent. In "Genomics and Health in the Developing World.' Edited by Kumar D. Oxford University Press, Oxford. 2012. pp 923-26.
[45] Rajab A, Bappal B, Al-Shaikh H, Al-Khusaibi S, Mohammed AJ. (2005) Common Autosomal Recessive Diseases in Oman Derived from a Hospital-Based Registry. Community Genetics (8):27-30.

Box 3.9 Clustered distribution of single gene disorders in GenTEE countries

(i) Geographical clusters

Country	disorder
Argentina	Sandhoff disease *(Cordoba province)* Oculocutaneous albinism *(La Rioja province)*
Brazil[2]	Albinism, undefined type *(Ilha dos Lençois region)* Achondrogenesis-Grebe *(southern Bahia region)* Acheiropodia *(southeast Minas Gerais region)* Spastic Paraplegia, optic atrophy and neuropathy - SPOAN – MIM 609541 (*Serrinha dos Pintos – Rio Grande do Norte – northeast Brazil*) Neural tube closure defects (NTD) and hydrocephaly not related to NTD (*São José do Pantano – Minas Gerais*) Gaucher Disease (*Tabuleiro do Norte – Ceará*) Mucopolysaccharidosis type VI, Phenylketonuria, Congenital Hypothyroidism and Cystic fibrosis (*Monte Santo – Bahia*)
Oman[6]	Congenital adrenal hyperplasia, galactosialidosis, Robinow syndrome *(Al Batinah)* Bardet-Biedl syndrome, Congenital generalized lipodystrophy *(Al Dakhiliyah)* Carbohydrate-deficient glycoprotein syndrome *(Al Dhahirah)* Ellis-van Creveld syndrome *(Al Sharqiyah)* Meckel-Gruber syndrome *(Al Wusta)* Schwartz-Jampel syndrome *(Muscat)*

(ii) Geographical and ethnic clusters

Country	Disorder
China	G6PD deficiency *(Dai, Jinuo, Yao and Zhuang minorities that live in autonomous regions in the Yunnan, Guangxi and Guizhou Provinces)*[3] G6PD deficiency and thalassaemia exhibit huge geographical variation. The China Ministry of Health (MOH) has not published national birth prevalence or prevalence of these disorders. The prevalence is much higher in southern China (Guangxi, Hainan, Yunnan, Guangdong and Guizhou province). Many studies have shown the carrier frequency of G6PD deficiency and thalassaemia are 5% or more in southern China (Table 4). In some areas the carrier frequency can exceed 20%. Due to extensive geographic size and variety of ethnic backgrounds, birth prevalence varies from the northern to southern areas of the country. Thalassaemia and G6PD are the most common genetic conditions in the provinces of Guangdong and Guangxi on the south coast of China.[4]
India[5]	ß-thalassaemia and SCD (Central India and the tribal belts in the west east and south, especially in Madhya, Pradesh, Chattisgarh, South Gujarat, Maharashtra and Orissa) Van der Knaap Cystic megalencephaly (Aggarwal community north India) Spinocerebellar ataxia type 12 (Aggarwal community north India) Butyryl cholinesterase deficiency (Vysa community south India) Laron dwarfism (western India and Sindh area of Pakistan)

(iii) Ethnic clusters and prevalence in at-risk populations

Country	Disorder
South Africa[7]	Fanconi's anaemia *(Afrikaans 1/26,000, Black 1/40,000)* Polycystic kidney disease: autosomal recessive type *(Afrikaans 1/26,000)* Galactosemia (*Black 1/18,000)* Duchene muscular dystrophy[a] (*Indian 1/14,000)* Oculocutaneous albinism (*Black 1/3,900)* Cystic fibrosis (*White 1/3,000)* Tay Sachs disease (*Ashkenazi Jewish 1/3,000)* Spinal muscular atrophy (*Black 1/2,000)* Gaucher disease (*Ashkenazi Jewish 1/1,600*) Sickle cell anaemia (*Black immigrants <1/10,000)* Thalassaemia (*Greek (β), Indian (α and β) >1/10,000)* Porphyria *(Afrikaans 3/1,000)*

[1] Penchaszadeh V (2012) Genetic testing and services in Argentina. Journal of Community Genetics. Online first 19 April 2012. Available at http://www.springerlink.com/content/18m202145ht67351/ (accessed October 17, 2012)
[2] Horovitz DDG et al. (2012) Genetic services and testing in Brazil. Journal of Community Genetics. Online first 5 May 2012. Available at http://www.springerlink.com/content/jh6080mp8134141h/ (accessed October 17, 2012)
[3] Data provided by Nanbert Zhong, Peking University Center of Medical Genetics, Beijing, People's Republic of China.
[4] Zhao X, Zhong N (2012) Genetic services and testing in China. Manuscript submitted for publication. Journal of Community Genetics. (DOI: 10.1007/s12687-013-0144-2).
[5] Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.
[6] Data provided by Anna Rajab, Genetic Unit, Directorate General of Health Affairs, Ministry of Health, Muscat, Sultanate of Oman.
[7] Kromberg J et al. (2012) Genetic services and testing in South Africa. Journal of Community Genetics. Online first 19 June 2012. Available at http://www.springerlink.com/content/p10460g7542372h5/ (accessed October 17, 2012)

Birth prevalence of selected common chromosomal disorders

Most countries (***Argentina***, ***China***, ***Egypt***, ***India***, ***Oman*** and ***South Africa***) were able to provide national data based on regional studies on the prevalence of Down syndrome (**Box 3.10**). The highest prevalence at birth was found to be in ***Oman*** where 2.6 births with Down syndrome per 1,000 live births were reported from 2000 to 2008. For this time period, the ascertainment of Down syndrome in the Sultanate was almost complete. In 90% of cases, the cytogenetic diagnosis was performed within 6 months after birth. Based on a case-control study, advanced maternal age was identified as a significant risk factor for Down syndrome. Some 40% of mothers giving birth to Down syndrome children in ***Oman*** are younger mothers. Thus over 60% of infants with Down syndrome are born to mothers of advanced maternal age. Currently the high prevalence of Down syndrome is considered to reflect the longer reproductive period in older women due to limited use of family planning and birth spacing. Consanguineous marriages were higher among parents of Down syndrome children than in the Omani population in general. The identification of possible additional risk factors for Down syndrome in ***Oman*** is presently being studied.[46]

Box 3.10 Prevalence at birth of Down syndrome (GenTEE countries with best available national data)

Country	Disorder	Birth prevalence (live births)
Argentina	Down syndrome	1:510
China[2]	Down syndrome	1:550
Egypt[3]	Down syndrome	1:787
India[4]	Down syndrome	1:800
Oman[5]	Down syndrome	1:380
South Africa[6]	Down syndrome	1:525

[1] Campaña H, Pawluk MS, López Camelo JS (2010) ECLAMC Study Group. Birth prevalence of 27 selected congenital anomalies in 7 geographic regions of Argentina. Arch Arg Pediatr 108 (5):409-417.
[2] Zhu J (2011) Genetic counseling and birth defect prevention. Paper presented at the National conference on early pregnancy and prenatal screening and birth defect prevention, Kunming, Yunnan Province, China
[3] Ezzat S. El-Sobky and Solaf M. Elsayed (2004): Down syndrome in Egypt. The Egyptian Journal of Medical Human Genetics, Vol. 5, (2): 67-78.
[4] Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.
[5] Rajab A, Patton M (2012): Genetic Diseases in the Sultanate of Oman. In: Genomics and Health in the Developing World. Oxford monographs on medical genetics by Dhavendra Kumar. Chapter 54, pp. 678-693.
[6] Kromberg J et al. (2012): Genetic services and testing in South Africa. Journal of Community Genetics. Online first 19 June 2012.

[46] Rajab et al.: Down Syndrome in The Sultanate of Oman (in preparation).

Prevalence of "late-onset" disorders

There are no data available on the prevalence of hereditary "late-onset disorders" in the GenTEE countries. Only ***Brazil*** was able to access estimates published in the official cancer report by the National MoH (2006)[47], citing an estimate of 2,400 to 4,800 new cases per year for hereditary breast cancer and 1,250 to 2,500 new cases per year of hereditary non-polyposis coli cancer.

In conclusion

All the GenTEE countries have limited available empirical epidemiological data. Several reasons underpin this problem including:

- poor or absent congenital disorder surveillance,
- lack of congenital disorder registries,
- lack of clinical diagnostic capability of health care practitioners especially in primary healthcare where most of these children present,
- home births,
- lack of access to appropriate care and
- underreporting.

This will limit any middle- and low-income country's ability to initiate and develop medical genetic services according to their health needs. Such data is essential to assessing those needs. The WHO recognized the inadequacy of epidemiological data in middle-and low-income countries, and its causes, as a barrier to developing medical genetic services in 1999.[48,49] It reiterated this in the recent *World Health Assembly resolution A63.17* in May 2010[50] that prioritized services for the care and prevention of congenital disorders, particularly in middle- and low-income nations. It further recommended that countries take steps to remediate the problem.

Acquiring such data in the short term will be difficult, time consuming and expensive. Some middle-and low-income countries may initially use modeled epidemiological data from the MoD *Global Report on Birth Defects* for health needs assessment for medical genetic services. Then part of the development of these services can be the acquisition of empirical epidemiological data. This approach was undertaken by the IR Iran in developing its medical genetic services.[51]

[47] Brazil (2006) Ministério da Saúde – Instituto Nacional do Câncer: Situação do câncer no Brasil. http://iah.iec.pa.gov.br/iah/fulltext/pc/monografias/ms/situcancerbrasil/situcancerbras2006.pdf (accessed may 16, 2013)
[48] WHO. Services for the prevention and management of genetic disorders and birth defects in developing countries. WHO, Geneva, Switzerland. 1999. [WHO/HGN/GL/WAOPBD/99.1]
[49] WHO. Primary health care approaches for the prevention and control of congenital and genetic disorders. WHO, Geneva, Switzerland. 2000. [WHO/HGN/WG/00.1]
[50] WHO. Birth Defects. 63rd World Health Assembly. WHO, Geneva Switzerland. May 2010. Online available at http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf (accessed May 3, 2013)
[51] Ashraf S: Genetic Epidemiology in Iran - a Basis for Service Development. PhD Thesis. University College London, 2009.

IV Availability of genetic services

Genetic service development

All GenTEE countries have embarked on the initiation and development of genetic services. Most countries have begun this process with limited or no empirical national data on the epidemiology of congenital and genetic disorders. **The concern is that informed service development may be hampered by the lack of data.**

"Early starters" like ***Argentina***, ***Brazil***, ***China*** and ***South Africa*** started genetic services in the 1950s and 1960s. Whereas ***the Philippines*** and **Oman** are relatively "late starters", initiating services in the 1980s and 1990s respectively. In ***Egypt*** and ***India*** services started in the 1970s[52]. Regardless of the onset, the **development of services was "champion driven"** in each GenTEE country. This means developed initially by a few physicians, many of them trained abroad.

Services in all countries were **started at tertiary care institutions, mostly at university based hospitals and academic departments and accompanied by the introduction of laboratory services**.

The **development of services was often funded by research means** or donation funds (and were at that time free-of-charge to the patient) and **depended on the priorities chosen by individual academics** acting in their country as early innovators and driving forces. Thus at the onset **genetic services development was fragmented**, characterized by *"enthusiasm based"* decision-making by individuals or institutions, resulting in unplanned service "silo" development.

Services are unregulated or regulations are often not enforced in some GenTEE countries as for instance in ***Argentina***, *"where DNA laboratories are certified by a state agency, participation in quality assessment programmes is voluntary and regulation very lax. Further, while in theory most of these laboratories must have in-house and external quality control programmes, there is little government oversight on these issues."*[53] (**Part 6**)

In some GenTEE countries, for instance in ***the Philippines***, genetic service development slowed down markedly when champions retired or died.

The diffusion of services into secondary and primary care has been top-down and dependent on the health care systems willingness to fund the necessary infrastructure and to pay for services. Thus in all the countries these services are limited, often resulting in fragmented public services.

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[52] First department of medical genetics established in India in Pune in 1972.

[53] Penchaszadeh VB, Aguiar MJB. (2009). Regulatory Issues in Clinical and Laboratory Genetics in Developing Countries. Examples from Latin America. In: Quality Issues in clinical genetic services. Eds Kristoffersson U, Cassiman JJ, Schmidtke J. Heidelberg: Springer Verlag, 2009.

In all countries the need for integrating genetic services into the public health care sector or strengthening public health care services was or still is not regarded as a health priority, resulting in **understaffed, underfunded and fragmented services in the public health sector and the lack of basic infrastructure facilities in peri-urban and rural areas.** However, recently some countries such as ***Argentina*** and ***Oman*** have started to implement national programmes in order to strengthen genetic services in the public health sector (**Box 4.1**).

Box 4.1 National policies to strengthen genetic services in GenTEE countries

In 1999 and the 2000s six out of eight GenTEE countries started to develop national policies to strengthen their genetic service infrastructure:

In 1999, the MoH in Oman developed a *"National Programme for the Control of Genetic Blood Disorders"* based on a community genetic model for controlling haemoglobin disorders by offering screening and counselling.
Since 2005, Oman has embarked on a systematic planned national development of genetic services outlined in the MoH's 5-year-plans.

In 2001, the National DoH in South Africa published *"Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities"*. The document provides recommendations for the integration of genetic services in South Africa. However, the effects on the health services imposed by the HIV/AIDS pandemic and other problems have severely limited the implementation of these guidelines.[1]

In 2002, the Egyptian MoH&P established a national committee for community genetics leading to the development of 11 genetic counselling clinics in different Egyptian governorates.

In 2006, Argentina appointed a national commission on genetic health services that has formulated a plan for strengthening genetic services in the public sector which was adapted by the National MoH as a national policy for strengthening the network of genetic services in the public sector and for supporting training activities in medical genetics addressed to primary health professionals in disadvantaged areas of the country. Recently, following the EU funded CAPABILITY project (2007-2009), special initiatives by national and provincial ministries of health have been started to improve genetic service delivery by increasing coordination and regionalization.

In 2009, the Brazilian National MoH published a decree which proposes the creation of a *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS). However, the decree is still awaiting implementation.

In 2010, the ***government of India*** established a National Institute of Biomedical Genomics in Kalyani, West Bengal.[2] This institute is likely to fill up the gaps in data on genetic disorders that exist at present. In February 2013, the *National Rural Health Mission of the Ministry of Health and Family Welfare (MoHFW)* launched a new initiative of *Child Health Screening and Early Intervention Services* to provide comprehensive screening and care to all children. The purpose of these services is to improve the overall quality of life of children through early detection of congenital disorders, diseases, deficiencies, development delays including disability. A set of thirty common conditions has been identified for screening and further management. An estimated 270 million children in the age group of 0-18 years are expected to be covered in a phased manner. Early intervention centres will be set up in all districts providing management for referred cases and will also link these cases with tertiary level health services in case of surgical management.[3,4]

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[1] www.doh.gov.za/docs/policy/humangenetics.pdf (accessed May 16, 2013)
[2] www.nibmg.ac.in (accessed May 16, 2013)
[3] http://www.newswala.com/Health-News/Launch-of-National-programme-for-Universal-Screening-of-Children-29206.html (accessed May 16, 2013)
[4] Ministry of Health and Family Welfare: Rashtriya Bal Swasthya karyakram launched - country's future depends on children's health. February 6, 2013. Online available at http://pib.nic.in/newsite/erelease.aspx?relid=92045 (accessed April 9, 2013)

Private sector development

Congenital and genetic disorders have not yet been recognized as a health priority by national health policy makers in any of the GenTEE countries. Due to this neglect, the private sector in several countries has stepped in during the last decade and increasingly provides genetic services not available in the public sector, and services covered or not covered by private health insurance or services for which long waiting times exist in the public health care sector.

The development of services in the private sector is opportunistic and mostly technology and market driven. Oft times testing services in the private sector are more or less unregulated and are offered regardless of their proven clinical validity or utility. Out-of-pocket expenses for services provided either by the commercial health sector (***Argentina***, ***Brazil***, ***Egypt***, ***India***, ***the Philippines***, ***South Africa***) or by government-owned services (such as ***China*** where for instance all genetic molecular tests provided by regional government owned secondary and tertiary care hospitals have to be paid for out-of-pocket) tend to be the norm for genetic services in most GenTEE countries, except in ***Oman*** where genetic services provided by the government are free for Omani nationals. However, in ***Oman***, being a late starter in the development of genetic services, its scope of currently available services is limited but steadily increasing.

Political and cultural influences on genetic service development

Although being "early starters" the development of genetic services was halted in ***Argentina*** and ***China*** by political developments. Many geneticists left ***Argentina*** in the mid-seventies because of the military dictatorship and in ***China***, when medical schools and universities were closed for 5 years, during the "Great Cultural Revolution" genetic services were suspended. In ***South Africa*** the further development of genetic services was halted in the late 2000s by the consequences of the HIV/AIDS epidemic.

Cultural traditions and population policies also shaped the development of services. In ***India*** and especially in ***China*** (due to the Chinese one child policy implemented in 1979), the establishment of PND services and related cytogenetic laboratories paved the way for implementing and diffusing genetic services into secondary and primary care. This development was supported by the willingness of the population to pay out-of-pocket for PND services.[54]

[54] In China, PND services are affordable to many Chinese couples *because „6 persons (4 grandparents, 2 parents) cover the cost for one child"* (personal communication N. Zhong, Director, Peking University Center of Medical Genetics, Peking University Health Science Center, Beijing, P. R. China)

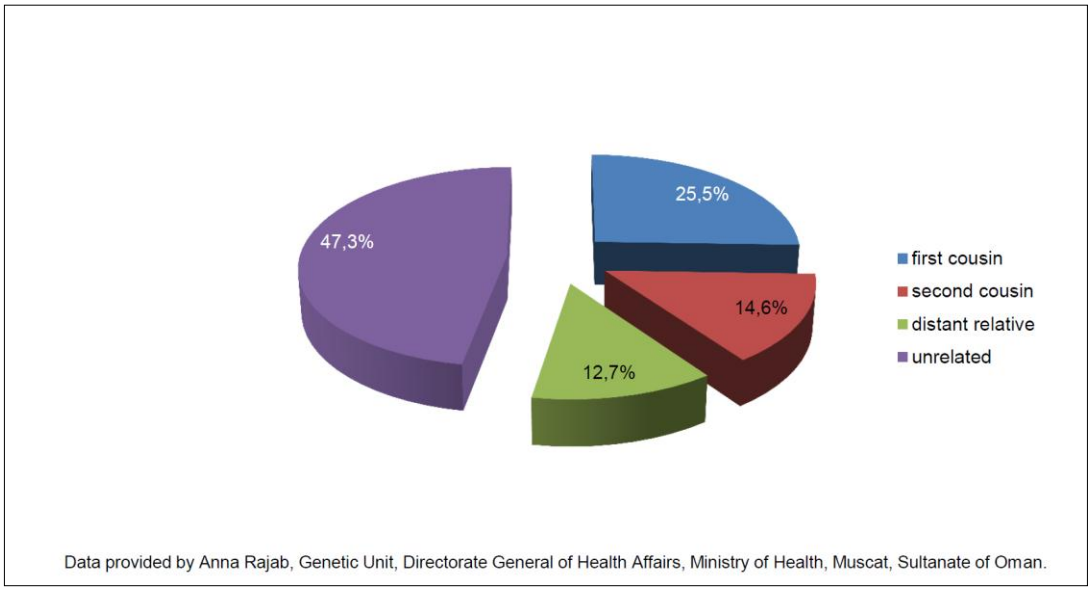
In countries with a strong Catholic tradition, such as ***Argentina***, ***Brazil*** and ***the Philippines***, where termination of pregnancy after a positive PND result is not legally permitted, the availability of PND and PGD services is very limited in the public sector. However, in these countries the private sector often steps in and offers services that are not funded by the state or by insurances and also provides services that are not strictly legal (see “Availability of key genetic services”).

In Muslim countries like ***Oman*** and ***Egypt*** the relative high frequency of rare single gene disorders due to consanguineous marriages, tribal structures of the rural population and the high prevalence of haemoglobin disorders furthered the development of genetic services including counselling services and establishment of hospital-based data registries.

Oman responded so far to this marriage pattern, deeply rooted in its culture, by offering voluntary premarital counselling and premarital screening services for haemoglobin disorders with current 10% uptake.
In 1999 the *“National Programme for the Control of Genetic Blood Disorders”* was established as a community genetic programme. It operates in all regions through regional teams providing care, high-risk population screening, premarital counselling and education (primarily for haemoglobin disorders).

In a recent population based survey on morbidity and mortality, funded by the ***Oman*** Research Council (TRC), the percentage of consanguinity (including first cousin/second cousin marriages) was 40.4%. When marriages among more distant relatives (marriages within same tribe) were included, consanguinity was well over 50% (53.1%) (**Figure 4.1**).

Figure 4.1 Consanguinity in Oman



Data provided by Anna Rajab, Genetic Unit, Directorate General of Health Affairs, Ministry of Health, Muscat, Sultanate of Oman.

In ***Egypt*** a recent hospital-based study showed that among those cases diagnosed with congenital or genetic disorders the overall parental consanguinity rate was 55%.[55]

In south ***India*** Hindus practice consanguinity (30 to 36 %), with preferred marriage being between uncle-niece. However in the rest of ***India*** the Hindus do not marry consanguineously. Whereas Muslims throughout ***India*** marry consanguineously. A multinational study of over 600,000 pregnancies and live births, in which 10 of the 38 populations studied were from ***India*** and nine were from Pakistan). The analysis showed that, from approximately the sixth month of pregnancy to a median age of 10 years, deaths in first-cousin progeny exceeded mortality in nonconsanguineous progeny by an average of 44/1000 births.[56,57]

Milestones in genetic service development in GenTEE countries

Table 4.1 provides a condensed overview of the service development during the last 60+ years. Service development in GenTEE countries is accompanied by

- professionalization of genetics through the establishment of professional bodies and scientific societies' development of qualification standards;

- recognition of medical genetics as a medical specialty in seven out of eight countries;

- increasing number of genetic units and available testing services primarily in the private sector or at tertiary care level;

- development of national policies for strengthening genetic services in the public domain in five (***Argentina***, ***Brazil***, ***Egypt***, ***Oman*** and ***South Africa***) out of eight countries (however, policies not yet implemented in ***Brazil*** or severely limited in ***South Africa***).

[55] Afifi HH et al.: The most encountered groups of genetic disorders in Giza Governorate, Egypt. Bratisl Lek Listy 2010; 111 (2), 62-69.
[56] Bittles AH. The impact of consangunity on the Indian population. Indian J Human Genet 2002; 8:45-51.
[57] Bittles AH. and Neel JV. 1994 The costs of human inbreeding and their implications for variations at the DNA level. Nature Genet. 8, 117–121.

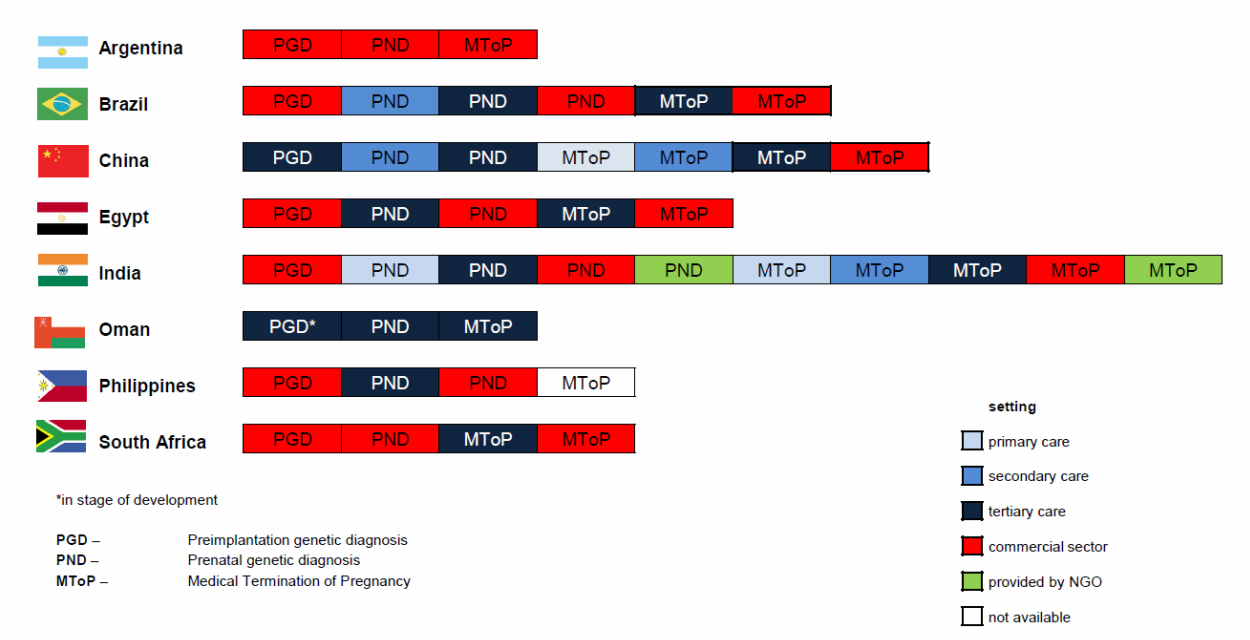
Table 4.1 Milestones in genetic service development in GenTEE countries

1950s/1960s	1970s	1980s	1990s	2000s	2010s
Argentina: Clinical genetic and cytogenetic services developed in a few public hospitals.	**Argentina:** National Centre of Medical Genetics established				**Argentina:** 80 clinical geneticists certified by the Argentine Society of Genetics staff approximately 41 clinical genetics units.
Brazil: 1950s: Brazilian Society of Genetics founded. 1960s: Clinical genetic and cytogenetic services developed in a few public hospitals.	**Brazil:** creation of ECLAMC (birth defects monitoring – research group); First medical residency programme in clinical genetics	**Brazil:** Brazilian Society of Clinical Genetics created. National database in Health (DATASUS) implemented	**Brazil:** Teratogen Information System	**Brazil:** Field for birth defect registry included in newborn declaration; National programme of newborn screening; Mandatory folic acid fortification in flour; National Policy for Comprehensive Care in Clinical Genetics published.	**Brazil:** 66 public genetic units, (47 clinical); approximately 50 private labs
China: First services started incl. cytogenetic diagnoses.	**China:** Informal genetic counselling services started in Hunan Province. Many genetic labs established. First molecular diagnostic lab established (Shanghai).	**China:** Molecular diagnosis for Thalassemia, DMD, Hemophilia A, etc. were developed based with RFLP, Southern blot, and PCR, on a research basis, in major academic institutions (Beijing, Shanghai, etc.).	**China:** 2nd national conference for medical genetics; tests are available for over 20 genetic disorders. Prenatal diagnosis offered widely.	**China:** Guidelines, regulations and specifications published to standardize medical genetic services are starting to be promoted by the government widely	**China:** A minimal 9 university bound medical genetic centres, 75 public labs
	Egypt: Centre of excellence in human genetics established in NRC.	**Egypt:** Centre for research and treatment of genetic diseases, ASU, established.		**Egypt:** National committee for community genetic services established by MOH&P.	**Egypt:** 11 community genetic clinics, 5 genetic departments at university/NRC level; 8 public labs, 10 private labs
	India: First genetic unit Department of Pediatrics, All India Institute of Medical Science, five other units develop subsequently. Institute of Genetics and Hospital for Genetic Diseases started in Hyderabad 1970. Indian Society of Human Genetics formed in 1971.	**India:** Sanjay Gandhi Post-graduate Medical Institute first department offering advanced courses in genetics both for physicians and scientists. 1st Newborn screening study in Bangalore (100,000 newborns screened for amino-acids disorders and G6PD deficiency.) Task Forces on human genetic disorders started by Indian Council of Medical Research (Mental retardation, Genetic counseling, Introducing genetic services into primary health care)	**India:** Molecular genetic studies in thalassemia and Duchenne muscular dystrophy established. First prenatal diagnosis of thalassemia made in Delhi using molecular techniques. Indian Human Genetic Variation project completed	**India:** National Institute of Biomedical genomics started in Kolkatta. Participates in International Cancer Genome Project (Oral ca). Ten centers providing training in medical genetics.	**India:** 50 genetic counseling centers, 10 public labs, 35 private labs in genetic tests (Biochemical, molecular, Cytogenetic)
			Oman: Pediatric genetic services commenced as individual efforts of Paediatric Consultants. National Committee for the Control of Genetic Blood Disorders & National Programme for the Control of Genetic Blood Disorders established.	**Oman:** Cytogenetic laboratory established in year 2000, Molecular genetic laboratory for hemoglobin disorders established; in 2006. Since 2005 planned national development of genetic services outlined in the MOH's 5-year-plans included community prevention of genetic diseases programme and training of nationals in modern genetic technology. National premarital screening service for haemoglobin disorders established. Sultan Qabus University Genetic Department & Research laboratories	**Oman:** 2 Genetic Units, 2 Cytogenetic labs 4 Molecular genetic labs 1 Metabolic lab at Sultan Qabus University National Genetic Center to open in 2012 National Cytogenetic Service Molecular genetic service Neonatal screening for Hypothyroidism Quality assurance from Eurogentest for Cytogenetic Services. Molecular laboratory for cancer care established; Oman Research Counsul gives priority to research in genetics
		Philippines: Genetic laboratory services and genetic counselling services introduced.	**Philippines:** Medical genetic unit established at UP-NIH; Dysmorphology clinic opened; Cytogenetic services opened; labs established; molecular genetics labs opened at UP-NIH. Medical Genetics Unit became the Institute of Human Genetics of NIH	**Philippines:** Biochemical genetic lab opened at the NIH; newborn screening programme implemented Offering of a Fellowship in Clinical Genetics in the Department of Pediatrics, UP College of Medicine	**Philippines:** 4 genetic units, 4 NBS (4 public), 1 Metabolic (1 public), 3 Molecular (2 public, 2 private), 1 Biochemical (1 public), 4 Cytogenetics (2 public, 2 private), 4 DNA analysis /paternity testing (3 public, 1 private) Offering of MS in Genetic Counseling at the Department of Pediatrics UP College of Medicine Philippine Genome Center launched
South Africa: Counselling services started at WITS and University of Cape Town; initiation of small cytogenetic labs	**South Africa:** DOH & W appoints Director of genetic services to set up community genetic services; first two academic chairs of Human Genetics; establishment medical genetic services; MOH genetic services declared integral part of health system funded by State.	**South Africa:** PhD in Human Genetics established; molecular laboratory established; new genetic test offered nationally; Master Degree Genetic Counselling	**South Africa:** Genetic counsellors registered; subspeciality of medical genetics recognised; basic clinical genetic services incorporated into PHC	**South Africa:** Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities published by DOH; development of services start to slow down due to the HIV/AIDS epidemic.	**South Africa:** 4 genetic units, 8 public labs, 5 private labs Public genetic lab services threatened by underfunding

Availability of key genetic services

Availability of PGD and PND services and follow-up services

Figure 4.2 Availability of PGD and PND services and follow-up services (2011)



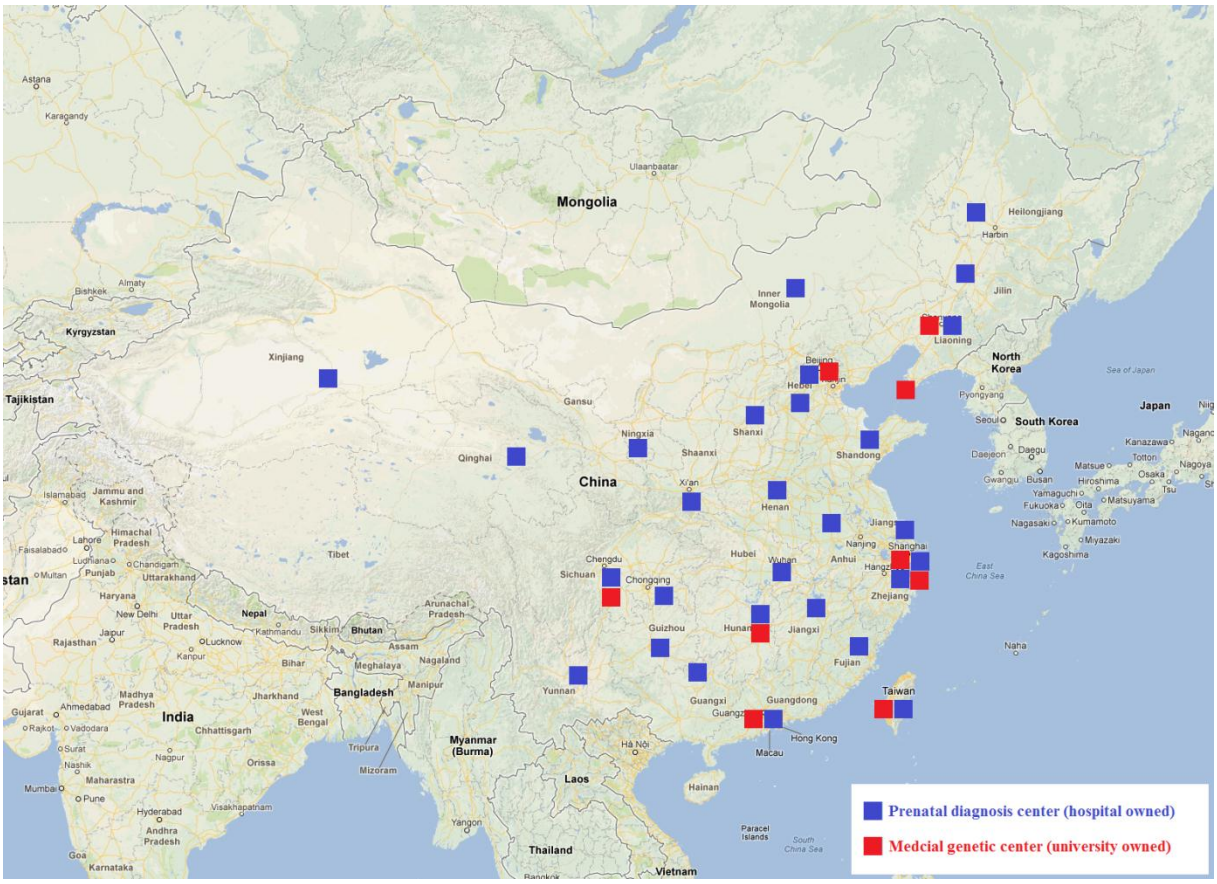
In ***Argentina***, reproductive genetic service and MToP are not available in the public domain due to the illegality of abortion. This legal restriction has prevented the development of PND, PGD and MToP services in the Argentinian public domain. However, PND and MToP are widely practiced in the private sector and seem to be "tolerated" by the system (**Figure 4.2**).

In ***Brazil*** – although abortion is illegal – court orders allow MToP in cases of severe congenital malformations[58], in particular if incompatible with life. PND services for monitoring pregnancies at risk have been established in some public settings. However, PND is most easily accessible in the private sector in ***Brazil*** where PGD is available as well.

In ***China*** and ***India***, prenatal genetic diagnosis paved the way for genetic service development. The geographical distribution of PND centres in ***China*** shows a distinct pattern (**Figure 4.3**). PND services diffused into secondary and tertiary care services and are more widespread than other medical genetic testing services. PND services cluster in the more affluent eastern and southern-eastern regions of ***China***. In the poorer western and northern region, less medical genetic services are available. But if services are available they tend to be PND services.

[58] In cases of anencephaly there is now a decision from the supreme court allowing pregnancy termination. (Horovitz D et al (2012) Genetic services and testing in Brazil. J Community Genet. Online available at http://link.springer.com/content/pdf/10.1007%2Fs12687-012-0096-y (accessed February 15, 2013)

Figure 4.3 The geographical location of medical genetic centres and PND centres in China



Overview provided by Zhong N, Zhao X: Peking University Center of Medical Genetics, Beijing, People's Republic of China.

In ***India***, PND is available in all care settings and ***India*** is the only GenTEE country where PND services are also provided by non-governmental organisations (NGOs).

The Philippines is the only country where MToP services are not provided not even by the private sector.

In all countries with PGD services, PGD services are only available in the private sector or, as in ***China***, PGD is provided by "entrepreneurial" hospitals owned by the government/regions and has to be paid for out-of-pocket.

In ***Oman***, PGD is in the first stages of development.

Availability of newborn screening services

Mandatory national newborn screening programmes have been implemented in all but one GenTEE country **(Figure 4.4)**. ***South Africa*** still has no national newborn screening programme and all newborn screening tests in ***South Africa*** have to be purchased in the private sector.

In ***India*** newborn screening is being provided by a number of government hospitals as well as private hospitals and stand-alone laboratories. The state of Goa has had a newborn screening in all the government hospitals for three years. In Gujarat newborn screening has recently been started in some government hospitals. Newborn screening for SCD has been initiated in Gujarat, Chattisgarh and Maharashtra both in tribal and non-tribal groups. A cohort of affected babies is followed up regularly and given early intervention care. The newborn screening programme has resulted in greater risk awareness among the parents of homozygous babies who are opting for PND in subsequent pregnancies.

China and ***India*** are the only GenTEE countries where NGOs provide newborn screening services in some areas. This is an indicator that they are covering service needs not provided by the state.

Figure 4.4 Availability of newborn screening services (2011)

However, the scope of disorders covered by national screening programmes differs substantially among countries and within countries (especially in ***Brazil***, ***China***; **Table 4.2**). ***Argentina's*** programme covers 10 disorders whereas the Omani programme, so far, covers only one disorder.

In ***the Philippines*** there are plans to expand newborn screening by 2013. Coverage will extend from 5 disorders to more than 20 disorders, including haemoglobin disorders, amino acid disorders, organic acid disorders, and endocrine disorders.

Table 4.2 Disorders for which newborns are screened in GenTEE countries[59] by national programmes (2011)

Country	Biotin. Def.	CAH	CF	CH	Chagas	Congenital deafness	Congenital syphilis	Galactosemia	G6PD	PKU	Retinopathy (preterm)	SCA	Thal.
Argentina	●	●	●	●	●	●	●	●		●	●		
Brazil			●[2]	●[3]						●		●[4]	
China[5]				●				●	●[6]	●			●[5]
Egypt[7]		●		●				●		●			
India[9]		●		●		●[8]	●		●			●[10]	
Oman				●									
Philippines		●		●				●	●	●			

Abbrevations:

Biotin Def.	Biotin Deficiency
CAH	Congenital adrenal hyperplasia
CF	Cystic fibrosis
CH	Congenital hypothyroidism
G6PD	Glucose-6-Phosphate Dehydrogenase deficiency
PKU	Phenylketonuria
SCA	Sickle cell anaemia
Thal.	Thalassaemia

1 No national/regional newborn screening programme available in South Africa.
2 CF is only screened for in 3 states
3 CH is only screened for in 27 states
4 SCA is only screened for in 14 states
5 Not available in Tibet.
6 Screened for in southern China
7 Newborn screening for PKU and Galactosemia are still in pilot testing phase
8 High risk newborns
9 For Biotin deficiency, cystic fibrosis, galactosemia and PKU available in private hospitals, and on demand in commercial sector.
10 In Gujarat, Chattisgarh and Maharashtra both in tribal and non-tribal groups.

[59] No national/regional newborn screening programme available in South Africa.

Coverage of national newborn screening programmes differs also between urban and rural areas. For instance, in ***Argentina***, in one third of jurisdictions providing newborn screening, coverage is less than 50%.[60] The highest coverage (close to 100%) occurs in the City of Buenos Aires.

In ***China***, the national wide newborn screening programme covers more than 90% of the newborn population in the affluent eastern provinces, falls well below 30% in the western provinces and is not available in Tibet. The national coverage in 2007 was 40%.[61]

In ***Egypt***, where newborn screening for CH was implemented as a preventive public health programme in 2000, screening for CH is available in all 29 governorates (>90% coverage).

In ***India*** many hospitals provide newborn screening for hypothyroidism, CAH and G6PD deficiency, while some hospitals provide further tests using tandem mass spectrometry (TANDEM MS). Screening for biotinidase deficiency, galactosemia, PKU and CF are available in commercial laboratories.

The mandatory national newborn screening programme in ***the Philippines*** started in 2004 with the *"Newborn Screening Act"*. Based on this act, newborn screening evolved from 24 hospitals in 1996 to more than 3000 newborn screening facilities today being available throughout the country. As of the end of 2011 newborn screening had an overall coverage of 42%.

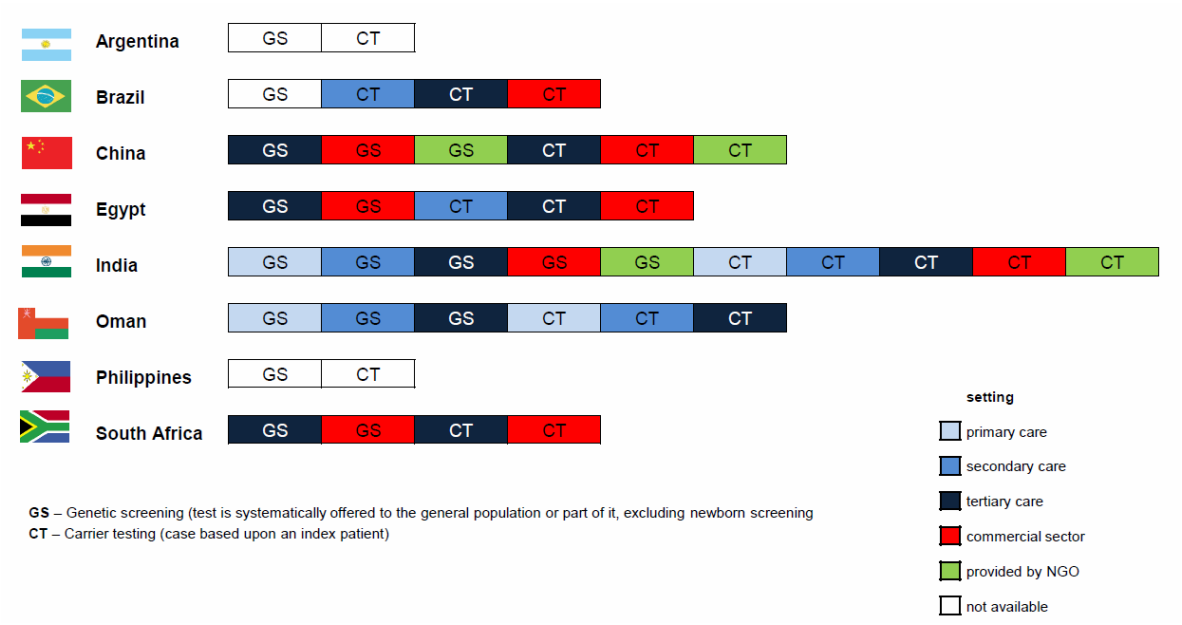
In most GenTEE countries with mandatory screening programmes, the private sector offers screening for additional disorders.

Availability of genetic screening[62] and carrier testing services[63]

It is mainly the Catholic GenTEE countries that do not have genetic screening and carrier testing.
Genetic screening tests are not available in ***Argentina***, ***Brazil*** and ***the Philippines***.
Carrier testing is not available in ***Argentina*** and ***the Philippines***.

[60] Argentina, 2009b. Ministerio de Salud, Departamento de Estadística e Información de Salud. Online available at http://www.deis.gov.ar/ (accessed May 16, 2013)
[61] Zhong N: Clinical Genetics in China: Genetic Disorders and Current Status. 2007, presentation available at www.capabilitynet.eu (accessed May 16, 2013)
[62] Newborn screening not included.,Screening is defined in the survey as a test that is systematically offered to the general population or part of it
[63] Heterozygote carrier for a autosomal-recessive condition

Figure 4.5 Availability of genetic screening and carrier testing services (2011)



In ***the Philippines***, most of the country's peoples are Roman Catholics, Protestants or Muslims, and in ***the Philippines***, **all these religions** oppose abortion for any purpose.

Brazil does not have genetic screening, however carrier testing is available in the private sector.

Conditions screened for:

In ***Brazil***, carrier screening is not performed on a population basis, but on a one on one basis, usually in the private sector when requested by the family or carrier. Carrier screening is performed in research projects linked to a specific disorders (e.g. in the region of Bahia for mucopolysaccharidosis VI cluster).

In ***India*** genetic screening services as well as carrier testing are available on request in the government hospitals, the commercial and private sector as well as not-for profit hospitals. Large scale screening for thalassaemia and SCD has been carried out in Gujarat by the Indian Red Cross Society from 2004 to 2010.[64] An ICMR initiated project has been completed in six cities with a high prevalence of haemoglobinopathies in six different states screening pregnant women and school children. [65] , [66] Although useful lessons how to implement carrier screening

[64] Indian Red Cross Society (IRCS), Gujarat State Branch. Annual Report 2009-2010. Ahmedabad; IRCS, Gujarat State Branch, 2010.
[65] Colah R, Surve R, Wadia M, Solanki P, Mayekar P, Thomas M et al.: Carrier screening for beta-thalassemia during pregnancy in India: a 7-year evaluation. Genet Test 2008; 12 : 181-5.

programmes have been learned from these studies, a coordinated national thalassaemia prevention programme has not yet been established in ***India***. However, plans are currently afoot to start screening and control programmes for thalassaemia in the high risk states including Delhi, Chandigarh and Mumbai[67] (see also **Box 3.6** above).

In ***Oman***: SCD, ß-thalassaemia and G6PD deficiency are screened for in the population. In the next 5-year plan it is expected that genetic screening will be introduced for more autosomal recessive disorders.

In ***South Africa***: In the private sector newborn screening is available and carrier screening for CF. In the tertiary health care sector there is carrier screening for individuals on request for numerous recessive disorders tested in the public health sector laboratory (includes CF, ß-thalassaemia, sickle cell anaemia, SMA etc.). Genetic screening for the Ashkenazi Jewish population (Tay-Sachs, CF, Canavan disease, FA, familial dysautonia, Niemann-Pick disease, Glycogen storage disease type 1a, Bloom syndrome, and mucolipidosis IV). Gaucher disease screening is available on request.

[66] Mohanty D, Colah R, Gorakshakar. Jai Vigyan S & T Mission Project on community control of thalassemia syndromes – Awareness, screening, genetic counselig and prevention. New Delhi: Indian Council of Medical Research; 2008.
[67] Verma IC, Saxena R, Kohli S: Past, present & future scenario of thalassaemic care & control in India. Indian J Med Res 134, October 2011, pp. 507-521.

Availability of genetic testing services[68]

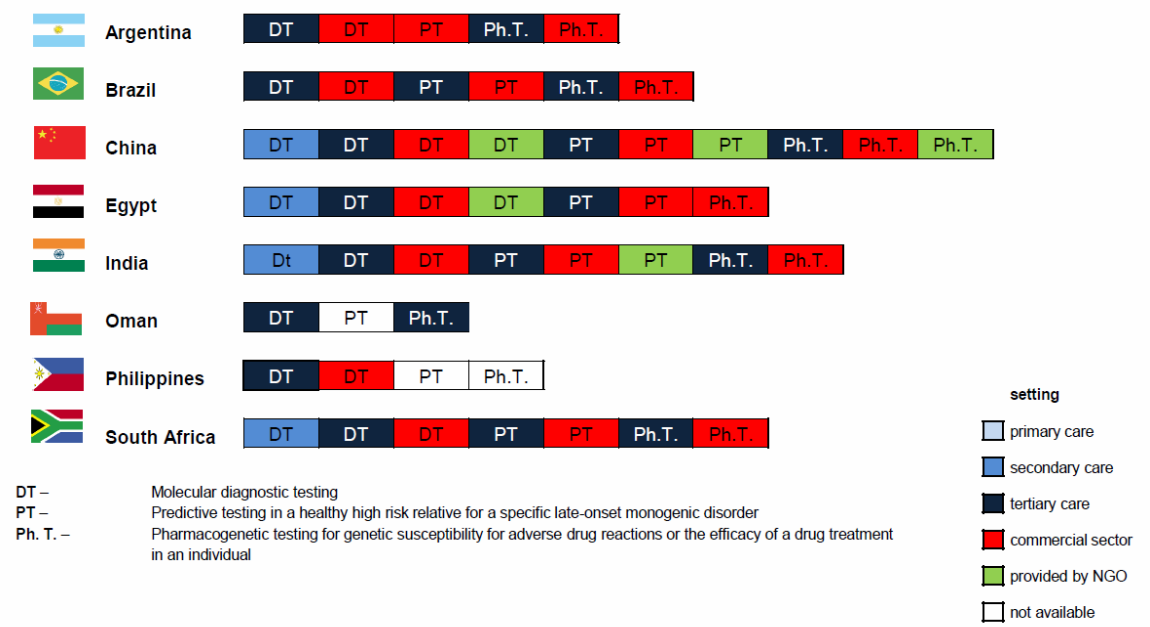
In all GenTEE countries molecular diagnostic testing is available at the tertiary care level (mostly university based). University based services are depending in part on research funds (especially in ***Brazil***, ***China*** and ***Egypt***) and may be free of charge and are limited by the availability of these funds.

In ***India*** molecular genetic testing is established and available for a large number of disorders varying from thalassaemia, Duchenne muscular dystrophy (DMD), SMA, fragile X syndrome, albinism, deafness, Wilson disease, Huntington disease (HD) and spinocerebellar ataxia and others. Many government sector laboratories offer molecular testing at subsidized rates. Laboratories in private institutions extend the range of disorders for which molecular testing is provided. Next generation sequencing (NGS) is available in a limited number of laboratories. Microarray analysis for cytogenetic studies are increasingly being used, being provided by several private laboratories and some government institutions. [69] A website (geneticsindia.org)[70] lists the genetic tests and services that are currently available in various laboratories in ***India***. The *Indian Genetic Disease Database* (IGDD) lists mutations observed in various genetic disorders.[71]

Figure 4.6 shows that genetic testing services are mainly available in the tertiary health care sector and the private sector. In ***China***, ***Egypt*** and ***India*** NGOs are offering testing services in order to facilitate access to diagnostic testing services in many underserved areas.

[68] Diagnostic testing in a symptomatic individual to confirm or exclude a genetic condition
[69] In India there are many cytogenetic testing laboratories country-wide, in secondary and tertiary care settings, and in government and the private sector. Many of them also provide PND, FISH studies and QF PCR analysis.
[70] http://www.geneticsindia.org/ (accessed April 12, 2013)
[71] http://www.igdd.iicb.res.in/IGDD/home.aspx (accessed April 10, 2013)

Figure 4.6 Availability of molecular genetic testing services (2011)



In ***Oman*** molecular diagnostic testing is provided free of charge by the MoH for: SCD, α-thalassaemia and ß-thalassaemia. Genetic tests for haematological cancer patients with tumour markers and minimal residual disease diagnostics are provided by the MoH's genetic laboratories.

In all GenTEE countries the basic testing required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available. Except for ***Oman*** and ***the Philippines*** this includes the ability to undertake predictive testing for late-onset monogenic disorders. Newer molecular genetic technologies such as microarray and NGS have been introduced for diagnostic service purposes in four countries (***Brazil***, ***China***, ***India*** and ***Oman***, **Table 6.3**, "Availability of different genetic testing techniques in GenTEE countries").

Pharmacogenetic testing has been introduced in all countries except ***the Philippines***.

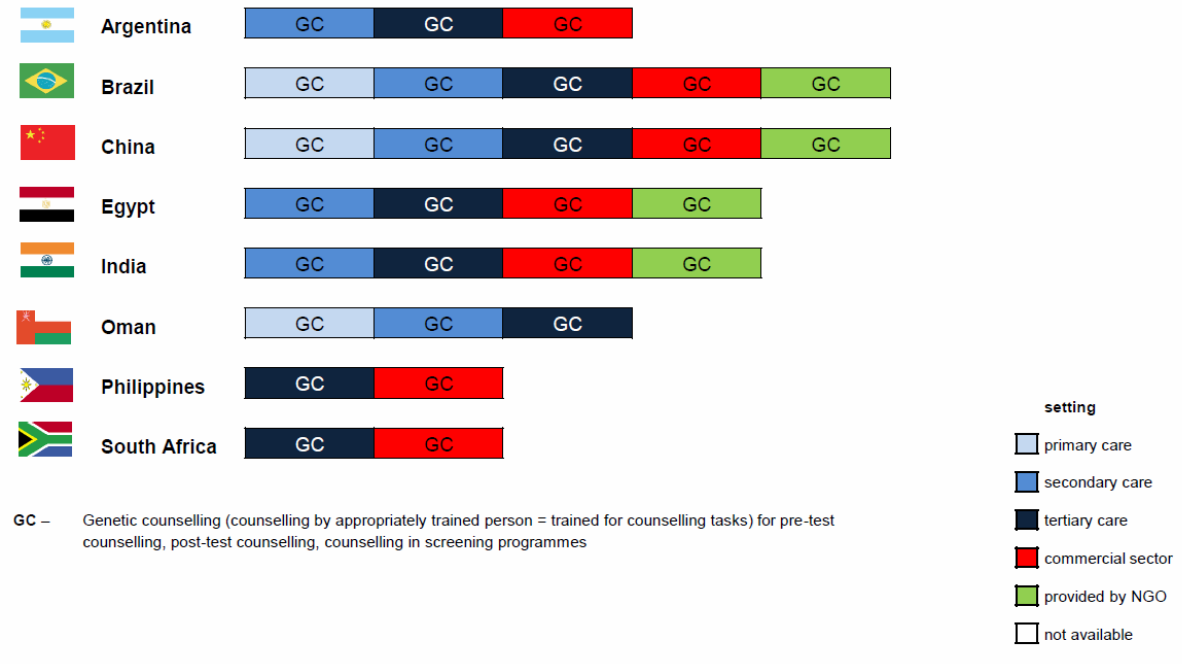
Molecular diagnostic testing for infectious diseases

Molecular diagnostic testing for infectious diseases is available in all the countries. This underlines the recognition and acceptance by the GenTEE countries of molecular genetic diagnostic testing in disciplines outside medical genetics.

Availability of genetic counselling services

Genetic counselling services have been established in all GenTEE countries **(Figure 4.7)**.

Figure 4.7 Availability of genetic counselling services (2011)



However, in ***the Philippines*** and ***South Africa*** genetic counselling services are exclusively available in tertiary care settings and in the private sector. This means that patients either need to travel to (mostly university based) urban counselling centres or must have the means to purchase genetic counselling in the private sector. Only three countries (***Brazil***, ***China*** and ***Oman***) provide counselling services at primary care level. However, in ***Brazil*** and ***China*** these services are only available in some regions and are not ubiquitous.

In four countries (***Brazil***, ***China***, ***Egypt*** and ***India***), NGOs provide genetic counselling services in some areas, indicating that they are covering counselling needs that are not met otherwise in these countries. In ***Brazil***, counselling provided by NGOs will only relate to a specific disease such as SCD. Usually in ***Brazil*** genetic counselling is performed by physicians in secondary or tertiary care settings.

Egypt (**Box 4.2**) and ***Oman*** have started to develop community genetic services based on counselling centres at primary, secondary and tertiary care.

In ***India***, genetic services were first established in Delhi, Pune, Hyderabad, Mumbai and Lucknow and included genetic counselling. When more genetic centres were set up in Bangalore, Chennai, Amritsar and other cities, they included genetic

counselling services as well. More recently the requirement for genetic counselling centres has been felt in the private sector, and a number of services have been set up in corporate hospitals.[72,73]

In ***South Africa*** in the 1990s an ambitious approach trying to offer medical genetic services including counselling services to the public through primary care was pioneered and policy guidelines established. However, implementing the guidelines and establishing a primary care counselling infrastructure was thwarted by the HIV/AIDS epidemic which forced the National Department of Health (DoH) to shift priorities.

Box 4.2 Development of counselling services: Egypt (2012)

In 2002 a national committee for community genetic services was established by the Egyptian government. Its main objective was to develop policy guidelines for the prevention and management of genetic disorders. The committee recommended the necessary establishment of a national programme for genetic services delivery. At the end of 2002 the Minister of Health responded to the request of the committee and signed an approval on a proposal presented by the MoH&P's Children with Special Needs Department that included an action plan and time frame for the implementation of a genetic counselling programme. The programme started 5 years ago with the establishment of one counselling clinic in Giza governorate.

Today (2012) there are 11 counselling clinics in different governorates in Egypt.

[72] Puri RD, Verma IC. Genetic services in India : A model for developing countries. In "Genomics and Health in the Developing World.' Edited by Kumar D. Oxford University Press, Oxford. 2012. pp 927-35.
[73] For profit proprietary hospitals in India generally owned by a corporate system.

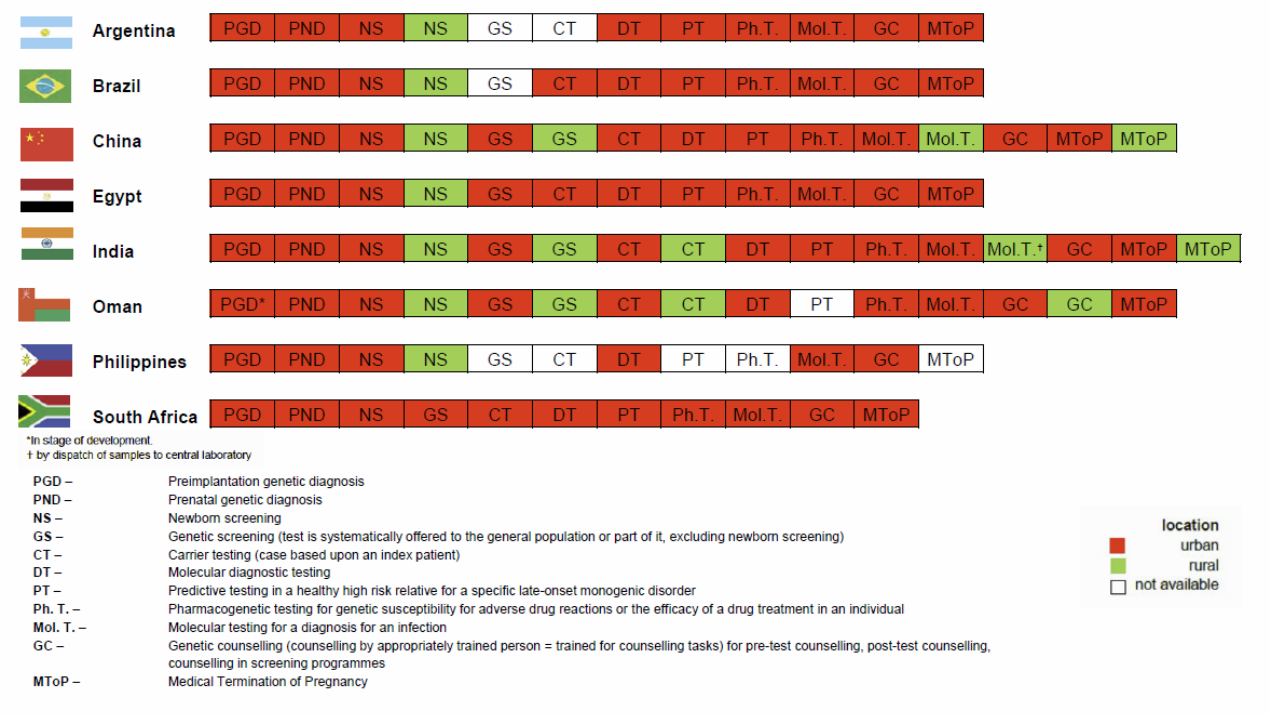
Availability of genetic testing services and follow-up services in urban/rural areas

Overall genetic testing services in the GenTEE countries are predominantly available in urban areas (**Figure 4.8**). In ***South Africa***, genetic services are exclusively available in urban areas. The current situation in ***South Africa*** is that most genetic services are either provided at urban tertiary care institutions, mostly universities, or in the private sector services that cater for the affluent urban upper middle and upper classes.

Figure 4.8 clearly indicates that the rural population is underserviced in most GenTEE countries and demonstrates the lack of basic infrastructure facilities in rural areas. In countries with national newborn screening programmes, services are also available in rural areas – however, coverage rates may differ between urban and rural areas. ***China*** and ***India*** are the only countries where MToP services are available in rural areas indicating the importance that is ascribed to PND services in these countries and the common practice of selective abortion.

In ***India*** private laboratories have extended the availability of genetic testing services, by appointing agents who collect the appropriate samples and send them onwards to the central laboratory for analysis. The expense for the service is borne by the patient.

Figure 4.8 Availability of genetic testing services and follow-up services in urban/rural areas (2011)



The geographical distribution of genetic services in ***Brazil*** (**Figure 4.9**) illustrates the disparities in the provision of services between urban and rural areas and between south, the eastern and north and western states. The majority of medical genetic services in ***Brazil*** are located in the south-east or in state capitals, the state of São Paulo being a noticeable exception, with services also available in the interior of the state. There are no genetic services in the states of Amazonas, Amapá, Roraima, Rondônia and Tocantins.

Figure 4.9 Geographical distribution of medical genetic services in Brazil (2010)

- Clinical care facilities in genetics
- Outpatient consultations in clinical genetics

Overview provided by Dafne Horovitz, Instituto Nacional de Saúde da Mulher, da Criança e do Adolescente Fernandes Figueira/Fundação Oswaldo Cruz, Rio de Janeiro, Brazil.

Availability of genetic services at different care levels

Genetic services in all GenTEE countries are dominantly available at tertiary care level and/or in private sector services concentrated in main cities. ***Oman*** provides community genetic services available in rural areas providing genetic screening and carrier testing services and genetic counselling primarily for haemoglobin disorders.

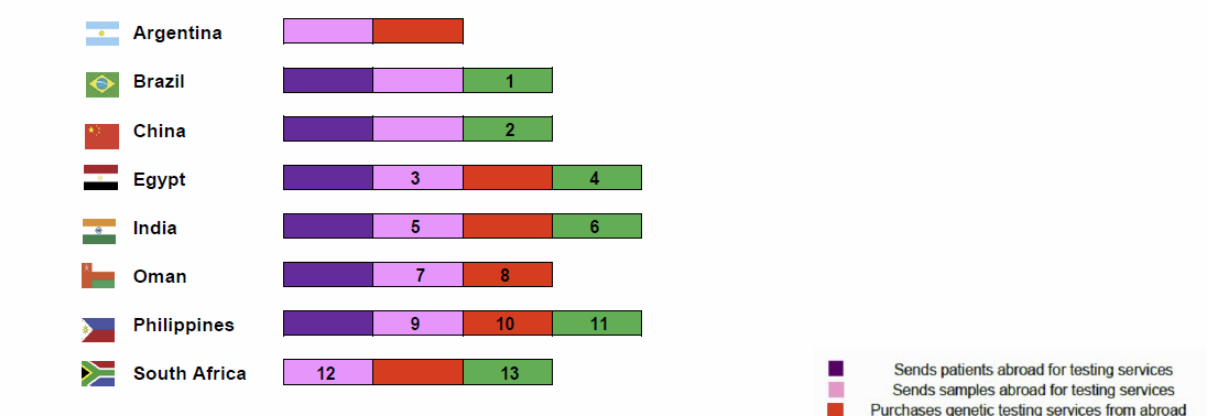
Patients or samples sent abroad/purchase of genetic testing services from abroad/provision of genetic testing services for foreign countries

The increasing trend of genetic tests crossing national boundaries can also be observed in GenTEE countries (**Figure 4.10**).

Purchasing genetic testing services from abroad – private sector dominated

Genetic services are not purchased from abroad by the public sector in most GenTEE countries (**Figure 4.10**). Only the ***Oman*** national health service purchases DNA based testing and biochemical testing services for mutation analysis for SMA, osteopetrosis, athrogryposis and TANDEM MS [74] screening for inborn errors of metabolism. This is changing as diagnostic capability increases in ***Oman***.

Figure 4.10 Patients or samples sent abroad/purchase of genetic testing services from abroad/provision of genetic testing services for foreign countries (2011)



1 For countries in South America, Africa and the Middle East
2 For South Asian countries
3 From Bioscientia/Germany
4 For patients from other Arab countries
5 Biochemical tests for diagnosis and prenatal diagnosis of Maple Syrup Urine Disorder and other biochemical disorders as well as molecular diagnosis
6 For neighbouring countries: Bangladesh, Pakistan, Sri Lanka, Maldives, United Arab Emirates and Iran
7 Mutation analysis for Spinal Muscular Atrophy, Osteopetrosis, Athrogryposis, Tandem MS screening for inborn errors of metabolism
8 For Huntington Disease, Duchenne Muscular Dystrophy
9 Molecular Genetics, Biochemical Genetics and Cytogenetics
10 Retinitis research Estonia
11 For Palau
12 Most samples are sent to the United Kingdom, the Netherlands or the USA.
13 For Central African Republic, Uganda and Kenya (sickle cell anaemia); Botswana, Namibia, Zimbabwe (chromosome analysis); for Lesotho for clinical evaluation, care, counselling and laboratory diagnostic services, as well as antenatal care

It is the private sector in most GenTEE countries (***Argentina***, ***Egypt***, ***India***[75], ***the Philippines***, ***South Africa***) that drives the trend to purchase testing services from abroad (**Box 4.3,** "Purchasing genetic testing services from abroad – the examples of Egypt and the Philippines").
In ***China*** and ***Egypt*** it is forbidden to send samples abroad for testing, exemptions need official approval from the government (**Box 4.3**).

[74] In Oman neonatal screening with Tandem MS is available in Sultan Qaboos University Hospital. Other tertiary hospitals purchase diagnostic services abroad.
[75] For molecular or biochemical tests not available in India, samples are sent abroad. For example, biochemical tests for diagnosis and prenatal diagnosis of Maple Syrup Urine disorder and some other biochemical disorders as well as molecular diagnosis of many diverse disorders.

The scope of testing services regularly obtained from abroad by the private sector in ***the Philippines*** exemplifies the extent to which the private sector uses the availability of overseas services. These services, which are easily accessible and increasingly becoming more competitively priced, are used to test for disorders that are not tested for within the country, thereby increasing the range of tests available.

Box 4.3 Purchasing genetic testing services from abroad – the examples of Egypt and the Philippines (2012)

Egypt

At a public authority level it is forbidden to send any human biological specimen abroad. It can be done sometimes for research purposes (with many restrictions) and for quality control. However, many private hospitals and laboratories send samples abroad for testing after getting official permission and obtaining the consent of the parents or patients for the following conditions: extended screening of newborns who were born in a private hospital, laboratory diagnostic and follow-up tests for metabolic conditions, chromosomal and genetic disorders, molecular testing for genetic disorders and diagnosis and predictive testing for some chronic illnesses including cancer. The most renowned external laboratory that private laboratories and hospitals regularly purchase genetic testing from is Bioscientia/Germany.

The Philippines

The private sector sends samples abroad for molecular, biochemical and cytogenetic testing for:

Molecular Genetics
α-thalassaemia carrier testing
Ataxia w/ oculomotor apraxia
Bullous Congenital
Ichthyosiform Erythroderma
Congenital Lipodystrophy
Cornelia de Lange Syndrome
Cystic Fibrosis
Fragile X carrier
Fragile X
Freeman-Sheldon Syndrome
Gaucher disease
Glycogen Storage Disease
Goltz Syndrome
PORCN gene analysis
Hereditary Spastic Parapesis
Holoprosencephaly
Huntington Disease
Hypercoagulable state
Lynch Syndrome
mtDNA disorder
Neonatal Diabetes
Leber's Hereditary Optic Neuropathy (LHON)
Pompe disease
Progeria
Rett Syndrome
XDP
XSCID

Cytogenetics
CGH-array
M-FISH
Microarray cytogenetics

Biochemical Genetics
Acylcarnitine and Amino Acid Profile (Expanded Newborn Screening) by Tandem Mass Spectrometry,
Acylcarnitine Profile for Fatty-Acid Oxidation Defects,
Amino Acid Quantification Analysis,
Biotinidase Screening,
7-Dehydrocholesterol Analysis,
Lysosomal Enzyme Assay,
DNA analysis of 6-Pyruvoyl Tetrahydropterin Synthase Deficient,
Hyperphenylalaninemia and Urine Pterins,
Transferrin Isoforms,
Phytanic Analysis,
Plasmalogen Analysis,
Very-Long Chain Fatty Acid for Peroxisomal Disorders,
Enzyme Assays for Lysosomal Storage Disorders [i.e. Gaucher's Disease, Niemann-Pick Disease, Pompe's Disease, Mucopolysaccharidosis (MPS) Enzyme Assay with Glycosaminoglycan (GAG) Screening].

Affluent patients who can afford the expense of travelling abroad to obtain genetic tests not available in their home countries are referred to other (mostly Western) countries for genetic testing services (e.g. PGD).

In ***Oman***, patients may be referred by the ***Oman*** national health services for PND services not available in the country. The national *"Treatment Abroad Expert Committee"* makes decisions on funding treatments and tests abroad for nationals in cases where such treatments and tests are unavailable locally.

Purchasing genetic testing from abroad

Although the private sector contributes to the global movement of patient samples in GenTEE countries and helps affluent patients to gain access to testing services, the quality of services received by affluent patients from abroad may be impaired due to the lack of consistent enforcement of quality controls in the private sector.

Testing for rare disorders[76]

Testing for rare disorders meets specific problems all over the world and no country is able to provide genetic testing for all such conditions. In the GenTEE countries testing for rare disorders is often only available from specialist research laboratories abroad and often the international community of senior scientists collaborates to provide the test and covers costs through research funds.

Providing testing services for other countries

Private laboratories in ***Brazil***, ***China***, ***Egypt***, ***India*** and ***South Africa*** are providing services for other countries, mostly for neighbouring countries with a less developed testing service infrastructure. ***India*** manufactures some kits for molecular testing and exports them.
Palau uses the national newborn screening programme in ***the Philippines***.

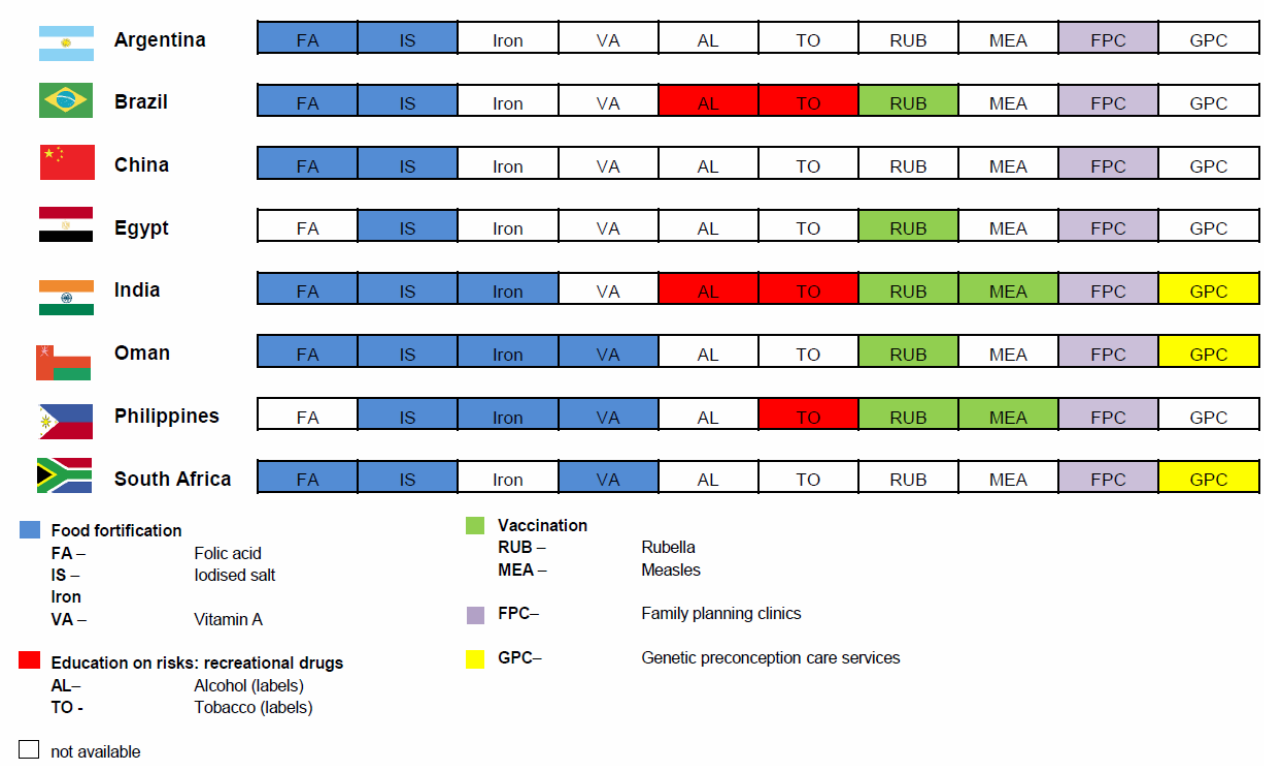
[76] Rare disorders are defined by the Council of the European Union as affecting no more than 5 per 10000 persons. „It is estimated that between 5 000 and 8 000 distinct rare diseases exist today, affecting between 6 % and 8 % of the population in the course of their lives." (Official Journal of the European Union: Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02). Available at http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF (accessed July 11, 2012)

Availability of national/regional preconception care programmes

Preconception care services available in the GenTEE countries are limited and are mainly community based services **(Figure 4.11)**. Family planning is available in all the GenTEE countries. Folic acid fortification of staple foods is available in 6 countries (***Egypt*** and ***the Philippines*** excluded) and iodised salt is available in all countries.

There is limited availability of iron (***Argentina***, ***Oman***, ***the Philippines***) and vitamin A (***Oman***, ***the Philippines***, ***South Africa***) food fortification. In ***India*** iodinisation of salt is mandatory and an ambitious programme has begun to administer iron and folic acid twice weekly to adolescent girls. Education regarding the adverse effects of alcohol and tobacco is limited to ***Argentina***, ***Brazil***, ***India*** and ***the Philippines***. Notably, ***South Africa***, with the highest documented community based birth prevalence of Foetal Alcohol syndrome has no universal education regarding alcohol. Rubella vaccination is not available in ***China*** and ***South Africa*** whereas in ***Oman*** vaccination coverage for measles and rubella is over 95%. Preconception care services for individuals, limited to the services described for public and private services for genetic counselling are available in ***India***, ***Oman*** and ***South Africa***.

Figure 4.11 Availability of preconception care (2011)



V Access to genetic services

Genetic services are highly inequitable in most GenTEE countries. The accessibility of available genetic services is compromised due to a number of inter-related barriers.

Cost and reimbursement systems for key genetic services

Coverage of genetic services by the public health care system or by compulsory social insurance or by private insurance is limited or often not available in GenTEE countries.

In ***Argentina***, patients with genetic disorders find it **very difficult to get insurance coverage for their conditions and usually have to go to court to obtain it**.
In ***India*** both social and private insurances **usually deny coverage of genetic services** on the grounds of "pre-existing condition".

In ***Brazil***, the majority of the population is served by the public Unified Health System ("Sistema Único de Saúde" or SUS) which proposes to ensure full, universal and free of charge medical access for the entire Brazilian population. However, genetic tests are often not available within the public health system due to insufficient number of services and scarce funding. **For those who have private insurance it has become progressively easier to have genetic testing covered.**

In ***China***, public hospitals provide most health care services which can be directly accessed by patients who know their way into the system.[77] Government-owned hospitals form the backbone of the health care system and account for most of the provision of health care services. With the start of economic reforms in ***China*** in 1978, the financial responsibility for providing health care services was decentralized. The responsibility for the provision of health care services shifted from the central national government to the local governments resulting in sharp inequalities between affluent urban areas and regions and poor rural areas/regions. Reducing government financial support meant that the government-owned providers in the health sector were forced to earn profit. Ownership of health services remained public but financing was gradually privatized, forcing health care facilities to rely more on the sale of profitable services. As a consequence, by the economic reforms government financial support was replaced by out-of-pocket spending (**see also Table 1.2**) and public hospitals came to function more like for-profit services[78] . There is comparatively little development of private hospitals and private hospitals are relatively rare.[79] However, backed by Western capital in the mid-2000s a private

[77] there are no GPs acting as gate keepers
[78] Blumenthal D and Hsiao W (2005): Privatization and Its Discontents — The Evolving Chinese Health Care System, New England Journal of Medicine 353;11, page 1166.
[79] „Currently, private hospitals are relatively rare, and private health care as an important component of the health care system in China has received little policy attention." (Huang C. et al: The Emerging Role of Private Health

medical sector has emerged to provide special services mainly in the Beijing, Shanghai and Guangzhou area. Health insurance coverage introduced in the 1990s has increased during the last decade. In 2005, 29% of the Chinese population were covered by a health insurance.[80] Since 2009, the Chinese central government has embarked on plans to provide universal coverage to all urban and rural residents by 2020. [81] Genetic counselling, prenatal screening, ultrasound examination, karyotyping, biochemical tests and treatment for congenital disorders may be to a certain extent covered by insurance. **However, most cytogenetic tests are not covered in *China*. Molecular genetic tests are new to the insurance authority and thus are exclusive.** Overall in ***China*** **personal affluence is a critical predictor of access to genetic testing services.**

In ***Egypt***, genetic services provided by the private sector have to be **covered mainly by out-of-pocket payments.** The coverage of genetic tests by the public sector is limited and services for poor people may be covered by donations from NGOs or individual charity.

In ***Oman***, the health system is characterized by its universal coverage for both citizens and governmental or public sector expatriates. The health system is financed mainly by the government and accounts for about 80% of the total health expenditure. Services purchased by private sector vary according to services. Health care is directly provided and operated by the government, with the MoH being the main health care provider. The MoH is also responsible for ensuring the availability of health policies and plans and monitoring the implementations. Although ***Oman*** has a growing health care sector, ***Oman*** still heavily depends on imports and there is a rising need for locally sourced health care products and services and locally trained health professionals. A large part of ***Oman's*** healthcare workforce comprises expatriates. This is changing due to the government's Omanization policy which encourages native doctors to pursue specialized courses outside the country.[82]
Genetic services are provided mostly by an Omani national workforce (95% Omanis).

Genetic services are free for Omani citizens (universal coverage), however, the scope of currently available testing services is limited. With the establishment of the National Genetic Centre, ***Oman*** aims at improving availability of and access to genetic services.

In ***the Philippines***, only newborn screening is covered by insurance, **all other genetic services and testing have to be paid for out-of-pocket.**

Care Provision in China. Asia Health Policy Program working paper series #10. Stanford University, Asia Health Policy Program, 2009: http://asiahealthpolicy.stanford.edu, accessed May 16, 2013)

[80] Blumenthal D and Hsiao W (2005): Privatization and Its Discontents — The Evolving Chinese Health Care System, New England Journal of Medicine 353;11, page 1167.

[81] WHO (2010): The world health report: health systems financing: the path to universal coverage.

[82] Sultanate of Oman Healthcare Report August 2012

In ***South Africa***, patients have to pay a fee for genetic testing services which is determined by a means test. Those patients, who access genetic services in private practice, generally have medical insurance and are reimbursed (depending on scheme and plan). For example, the cost of a genetic counselling session is set, approximately, at the accepted medical aid rate for such a consultation and therefore is largely refunded to the patient by the medical insurance.

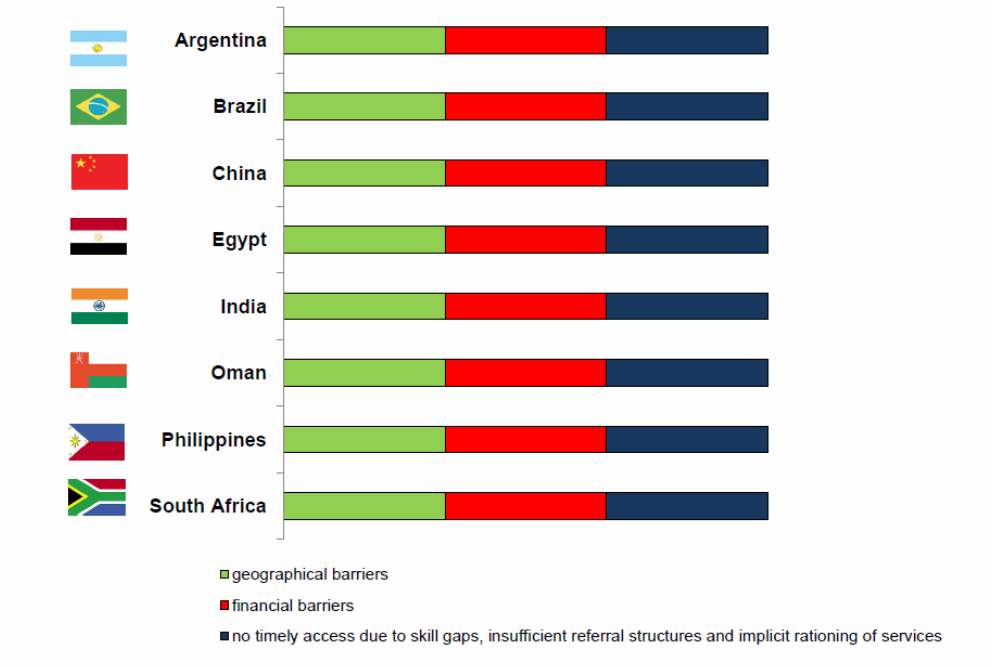
Coverage of genetic services

The GenTEE survey clearly indicates that the **unwillingness to cover genetic tests by both public health care and (private) insurance companies stalls the comprehensive clinical integration of genetic testing services into routine health care**. However it has to be kept in mind that coverage and reimbursement decisions worldwide take time and are often hampered by a lack of available data evaluating the cost-benefit of genetic tests and by the costs incurred by the evaluation of new technologies.

Significant barriers that affect access to medical genetic services in GenTEE countries

Although all factors shown below in **Figure 5.1**, **geographical barriers, financial barriers, no timely access due to skill gaps, insufficient referral structures and implicit rationing of services, operate in all GenTEE countries** the proportion of each is likely to vary. For example in ***India*** the financial barrier is greater than others. However it is beyond the scope of this survey to assess the contribution of each factor.

Figure 5.1 Factors reported to affect access to medical genetic services in GenTEE countries



Barriers that affect access to medical genetic services

- ***Geographical accessibility of services:*** In all GenTEE countries **genetic services are concentrated in the main cities, impacting the ability of the poorer rural and peri-urban population to physically reach the services**. Public transportation to services may be inadequate, transport costs may be prohibitive, the time it takes to undergo a consultation may take a day or more resulting in the individual patient/parents losing time from work and thus valuable pay. The distance for specimens to be transported may also be great resulting in time delays that can affect the viability of specimens, in particular specimens for cytogenetic analysis. In order to overcome geographical barriers, in some countries such as ***Argentina, India*** and ***the Philippines*** genetic centres in tertiary hospitals run telemedicine programmes for genetic consultations accessible to secondary and primary care hospitals located in distant regions. In ***India*** telemedicine is being introduced in certain regions and free educational telemedicine conferences are organized for medical geneticists.

- ***Affordability of services:*** **Out-of-pocket payments in the private sector tend to be the norm for genetic testing services, and mostly the affluent upper-middle and upper classes can afford services.** The major barriers to equitable access in most GenTEE countries are the lack of universal coverage and the dependence of genetic service provision on direct payments. **The majority of patients and families cannot afford out-of-pocket funding** in countries such as ***Argentina***, ***China***, ***Egypt***, ***India*** and ***the Philippines***.

- ***Adequacy of services:*** In most GenTEE contries **fragmented, underfunded and understaffed public health sector services are unable to deliver the volume of required services.** This results in crowded services, excessive waiting lists that implicitly lead to non-transparent prioritization and rationing of services. **All GenTEE countries are in need of better referral system development.** In GenTEE countries genetic services are available at specialized hospitals at secondary or tertiary care level and routine points of entry to genetic service at primary care level are very limited.[83] **Lack of expertise and skill gaps in recognizing congenital and genetic disorders by primary care providers impedes the route through necessary diagnostic, care and prevention services for patients and their families.** Community genetic services near to patients and their families throughout the country are rare and can only be found to a certain extent in ***Oman***, yet with restricted scope of services.

[83] Mandatory newborn screening programme may be the only exception for which in some countries exist entry points at primary care level.

VI Current state of genetic services

Human resources and training

Integration of medical genetics into the medical schools' curricula

In ***Argentina***, genetics knowledge of physicians is poor as most medical schools do not include meaningful teaching in genetics in their curricula.

In ***Brazil***, genetics is part of the curricula in several health related graduate schools, either as a structured discipline or among one of the major themes such as cell biology. It is classically taught within the basic disciplines of the courses with little, if any, integration with practice. Few medical school curricula include practical training in genetics. The genetics content in almost all medical schools does not cover even the needs of a general medical education. Therefore, most physicians do not recognize the genetic basis of diseases with which they are dealing and/or do not know how to refer to genetic services and/or do not give the deserved importance to the process of genetic counselling.

In ***China***, approximately 100 universities/colleges are qualified for giving 5-8 years professional education for the students wanting to be medical doctors (MDs). All these universities/colleges have genetics as compulsory, less than 10 universities, including Peking University, Tshinghua University and Zhejiang University, have specially designed medical genetics instead of genetics. None of these set up separate course for medical genetics. In ***China***, medical genetics or overall genetics usually is taught in the 2nd year of all medical undergraduate (including the courses for doctors, nurses, medical test technicians, public health personnel, medical administration personnel) as compulsory before the beginning of medical practice.“[84]

In ***Egypt*****,** medical genetics is integrated in the medical faculties' curricula for undergraduate students but not as a separate specialty. It is taught as part of the paediatric curriculum.[85]

In ***India***, genetics is presently taught under various specialities like anatomy, physiology, pathology, paediatric s and internal medicine, as there is no separate department of genetics in the majority of medical schools. Although the Medical Council of India (MCI) has incorporated medical genetics in the medical curriculum, most medical schools are considered ill-prepared for medical genetics.[86]

In ***Oman***, genetic courses are taught in genetic biotechnology and biology at Sultan Qaboos University (SQU) (Medical Laboratory Scientific Officers (MLSO) and College

[84] Zhong N. GenTEE Report on People's Republic of China, 2010, p. 21.
[85] Randa Kamal A. Raouf (2010): GenTEE Report on Egypt, p. 35.
[86] Aggarwal SS: Medical Genetics in India – What needs to be done? Indian J Med Res, 130, October 2009, pp. 354-356.

of Medicine and Health Science). Medical school courses include basics of human genetics, chromosomal and molecular inheritance. Clinical genetics are taught in 5th-7th year of medical paediatrics and internal medicine.

In ***the Philippines*****,** genetics is taught primarily in medical school as topics integrated in biochemistry, paediatrics, internal medicine and obstetrics (Commission on Higher Education (CHED) 2006). The *Philippine Paediatric Society* has included genetics as a core topic in its paediatrics curriculum for all medical schools.

In ***South Africa***, four medical schools in the country have medical genetics professionals on their staff, and medical genetics is integrated into the undergraduate student curricula to a varying extent. Medical students are also trained at three other universities, but medical genetics teaching at these universities is limited and often falls to clinicians in various non-genetic specialties, particularly paediatrics.

Impact of unsatisfactory integration of medical genetics into medical schools' curricula

Due to typically unsatisfactory integration of medical genetics into their undergraduate medical training, it is fair to assume that most physicians in GenTEE countries, do not recognize the genetic basis of diseases of their patients, do not know how to refer to genetic services, if available, and do not give due importance to genetic counselling.

Medical genetics recognised as a specialty in medicine

Although medical genetics is recognized as a specialty in all GenTEE countries but ***China***, GenTEE countries are underserved and the number of genetic specialists is insufficient in regard to the recommendations from from the UK *Clinical Genetics Committee of the Royal College of Physicians*[87] **(Table 6.1)**.

Table 6.1 Recognition of medical genetics, estimated numbers of certified medical geneticists (2011)

Country	Specialty officially recognized	Certified medical geneticists	Per million population
Argentina	1991	~120	2.8
Brazil	1983[88]/1993[89]	~200	1.0
China	recognised as a sub-specialty	-	-
Egypt	yes, date not provided	~ 100-150[90]	1.12-1.74
India	1982 (super specialty)[91]	60	0.06
Oman	2000[92]	5	2.5
Philippines	2000	7	0.07
South Africa	2007	11	0.22

***Argentina*:** The specialty of medical genetics is recognized by the National MoH since 1991. The *Argentine Society of Genetics*, in conjunction with the National MoH, has certified about 120 clinical geneticists who staff 41 clinical genetics units throughout the country or work in private practice, providing genetic consultations and counselling.

***Brazil*:** In 1983, medical genetics was recognized as a medical specialty by the *Conselho Federal de Medicina* (CFM, Federal Council of Medicine). Since then, several new residency programmes were created, totalling 11 programmes, and vacancies for 23 new physician trainees yearly. Expertise in a medical specialty in

[87] A report from the Clinical Genetics Committee of the Royal College of Physicians suggests that two medical geneticists and four genetic counsellors are required to provide an adequate genetic service to every one million people.(Harper PS, Hughes HE and Raeburn JA: Clinical genetics services in the 21st century. The Royal College of Physicians of London, London, 1996. Online available at http://www.rcplondon.ac.uk/sites/default/files/documents/clin_gen_21c.pdf (accessed May 6, 2013)
[88] Federal Council of Medicine
[89] Board Certification Brazilian Medical Association
[90] Data provided by Prof. Mona Mohamed Hassan El Ruby, Clinical Genetics, NRC, Egypt.
[91] Doctor of Medicine (DM) in Medical Genetics is a three-year super-specialty course offered only by a few medical colleges in India and only a single government -run medical college offers this course. http://entrance-exam.net/government-colleges-for-dm-in-medical-genetics/ (accessed April 12, 2013)
[92] Presently there are five clinical geneticists in Oman qualified in clinical genetics from UK and Canada. In genetic laboratories currently there are 5 PhD Scientists, 10 senior genetic scientists with master degree and 30 genetic laboratory scientists

Brazil is not based solely on the titles coming from medical residencies and recognized by the Ministry of Education. Another form of professional recognition comes from board certification, awarded by the societies of medical specialties and recognized by the *Associação Médica Brasileira* (AMB, Brazilian Medical Association) and the CFM. Board certifications in medical genetics are held annually since 1993 by *Sociedade Brasileira de Genética Médica* (SBGM) and involve a theoretical test, analysis of curriculum and interviews. In 2010, around 200 physicians had been awarded with board certificates.

China**:** Medical genetics is currently recognized as a sub-major in medical school but a sub-specialty in prenatal practice in the country. In most of cases, the medical genetics professional in medical school has nothing to do with clinical genetic services. Genetic testing and genetic counselling are run independently in hospital, and generally are not combined and considered as the term of "medical genetics". Clinical staffs who undertake the role of "medical geneticists" are normally obstetricians, gynaecologists, paediatricians, and various other medical specialties. In some small local genetic posts even genetic laboratory technicians may play the role of "genetic counsellor", due to the lack of professional staff, although it is illegal.

Egypt**:** Medical genetics is a recognized specialty in the country. The number of practicing medical geneticists is estimated to be 100-150.

India**:** Medical genetics is recognized as a super specialty but the MCI has not made it mandatory that every medical school should have a department of medical genetics for training purposes. There are only ten medical schools/institutions that provide postgraduate training in clinical genetics. There is only a single government-run institute in ***India*** offering a three-year postgraduate training course in medical genetics (DM in medical genetics[93], Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences[94]). However this institute has a MCI approved intake capacity of just two students per year. For seeking admission to the DM in medical genetics, candidates must have completed their Medical Doctor (MD) in paediatrics or general medicine or candidates with MD/ master of science (MSc) in obstetrics & gynaecology are also eligible to apply for this course. Postgraduate training in medical genetics is also being introduced in three more institutions. At least six other institutions provide training in medical genetics without a formal degree.

Oman recognizes a specialty in medical genetics obtained abroad.

In ***the Philippines*** there are only seven trained medical geneticists, five of whom are in Metro Manila while two are practicing in the provinces. To date, all clinical geneticists are paediatricians. Medical genetics is a recognized specialty since year

[93] http://entrance-exam.net/government-colleges-for-dm-in-medical-genetics/ (accessed April 12, 2013)
[94] http://www.sgpgi.ac.in (accessed April 11, 2013)

2000. The Department of Paediatrics, Philippine General Hospital (PGH) offers a 2-year fellowship programme in clinical genetics. It is designed to provide broad clinical exposure to areas of dysmorphology, biochemical genetics, cytogenetics, molecular genetics, and neonatal screening programmes. Components of the training programme are genetics and metabolic clinics, ward rounds and participation in regularly scheduled pre- and post-clinic conferences. Experience in genetic counselling and training in laboratory procedures for the diagnosis of genetic disorders are likewise included.[95]

South Africa*:** Medical genetics was initially recognised as a subspecialty, and in 1999 nine medical geneticists were registered, through a grandfather clause, under this system. Subsequently nine more medical specialists (mostly paediatricians) undertook the newly introduced 2-year medical genetics training and were registered as sub specialist medical geneticists. However, in 2007, medical genetics was recognised as a primary specialty in medicine in ***South Africa. Specialist training (over 4 years) towards a postgraduate medical degree and fellowship of the *College of Medical Geneticists* (FCMG(SA)) is currently offered at four universities. At present, there are 11 medical geneticists, registered with the *Health Professions Council of South Africa* (HPCSA), of which ten are in academic practice. Seven registrars are in specialist training.

Impact of limited availability of medical geneticists

Especially in ***Brazil, China, India, the Philippines*** and ***South Africa*** there is an urgent need for expansion and capacity building in medical genetics. The limitation in available medical geneticists not only severely hampers the ability of these countries to diagnose and manage hereditable disorders but also their ability to incorporate the benefits of genetic/genomics research into mainstream medicine.

[95] Padilla CD. Overview of Genetic Health Services at UP Manila. Acta Medica Philippina. 2008; 42 (2): 7-10.

Postgraduate training programmes available for biochemical, cytogenetic and molecular geneticists

***Argentina*:** There are postgraduate training programmes in biochemical, cytogenetic, and molecular genetics for PhD professionals, but these are highly variable in scope and the number of graduates unknown.

***Brazil*:** The postgraduate programmes in genetics have been collaborating with the training of professionals for teaching and research in medical genetics. Although the primary purpose is to train professionals for universities and research centres, a reasonable proportion become trained in the specific areas of medical genetic laboratory investigation including cytogenetics, biochemistry, and molecular biology. The non-medical professionals whose undergraduate or postgraduate training enables them to perform within medical genetics, or even those who enter the area by the practice, may obtain qualification through their associations, although few opt to do so.

***China*:** Postgraduate training programmes for biochemical, cytogenetic and molecular geneticists are not available.

***Egypt*:** No postgraduate degree for any of the genetic laboratory specialties exists, thus physicians receive either a master in genetics or in clinical pathology, while pharmacists and scientists receive a master degree in molecular biology or master of biochemical science. To become qualified as genetic laboratory specialists, they get extensive training for one year and then on-job training for a second year in any of the genetic laboratories in the university or the *National Research Center* (NRC).
There are also postgraduate specialized training courses in molecular genetics, cytogenetics or biochemical genetics that are done regularly twice per year, in the genetic division of the NRC.

India*:** Postgraduate training programmes for biochemical, cytogenetic and molecular geneticists are available in many universities. Many departments of biotechnology have been opened in universities, providing training in molecular genetics, but also biochemical and cytogenetics. Due to the numerical availability of laboratories in ***India, employment of trained personnel is assured and there is no shortage of scientists trained in molecular techniques. Biochemists are mostly pursuing molecular based research.

***Oman*:** Master degrees in molecular genetics, molecular biology, biotechnology, and biochemistry are available at the SQU. Professional training in cytogenetics has been provided at the national cytogenetic laboratory.
Molecular cytogenetics (FISH) training was provided by visiting consultants.
A PhD programme in biochemical and molecular genetics is available at SQU.

***The Philippines*:** Formal postgraduate training programmes for molecular genetics are available. A MSc in molecular biology and biotechnology and a PhD in molecular biology and biotechnology are currently offered at the University of the Philippines (UP) Diliman; a MSc in molecular medicine is currently offered in St. Luke's Medical Center (SLMC); at the UP Manila College of Medicine, there is a MD-PhD programme with a focus on PhD on molecular medicine. However, formal postgraduate training programmes for biochemical genetics and cytogenetics are not yet available. Collaborations with hospitals abroad are available for further training of clinical geneticists. In-house trainings and seminars are carried out by the faculty and senior laboratory scientists of the Institute of Human Genetics-National Institutes of Health (IHG-NIH).

***South Africa*:** Medical scientists can be trained in human genetics if they complete a bachelor of science (BSc) (Honours) degree in a biological science and a 2-year internship, in a recognised human genetics setting, and become registered by the HPCSA.[96]
Similarly, medical technologists need to complete a 3-year national diploma or BSc degree in biomedical technology and a 1-year internship, and then register with the HPCSA.
Medical scientists have the option of undertaking further study towards obtaining MSc or PhD degrees.
South Africa has training programmes sufficient to train the numbers needed to undertake the work required to develop medical genetic services. The problem lies with the national and provincial health departments lacking the will, commitment and finances to support this training and make posts available for those trained.

[96] Health Professionals Council of South Africa (2010). Professional boards. Medical technology. Available from: http://www.hpcsa.co.za/board_medtech_education.php (accessed May 16, 2013)

Availability of postgraduate programmes

With the exception of ***China*** and ***Egypt*** (where postgraduate training is more on a CME basis) postgraduate programmes for laboratory geneticists are available in the GenTEE countries. They are variable regarding scope and capacity, and limited to an extent in some, for example in ***the Philippines*** and ***South Africa***, by lack of resources. Countries have, and still do, send scientists overseas for training, but as a country's capacity grows, doing this, at least for basic training, has become less necessary.

The availability of postgraduate programmes is impacted by:

Lack of training infrastructures

In ***the Philippines***, there is a lack of diagnostic and treatment facilities. Added to this is the scarcity of geneticists who could oversee trainings and make consultations at the same time.

With all the developments in the field of genetics, especially in research, and the growing number of patients, it is more cost-effective for some countries to send people abroad for training (***Oman***, ***the Philippines***). However, ***Oman*** is in transition and is setting up more own training capacities.

Lack of political commitment and resources

South Africa has training programmes to train the necessary staff to undertake the work required to develop medical genetic services. The problem is the necessary national and provincial health departments' will, commitment and lack of finances to undertake this training and make posts available for those trained.

Genetic counsellors as a recognised and registered profession

Genetic counsellors are a registered profession in ***South Africa*** only**.**

In the other GenTEE countries genetic counselling is mostly provided by clinical geneticists and by various other medical professions:

Argentina**:** Non-physician genetic counsellors are not a recognized or registered profession, and there is no formal postgraduate training in genetic counselling. Genetic counselling is a physician responsibility and is provided by clinical geneticists or other physicians with genetics training.

Brazil**:** Despite the involvement of many professionals in care regarding genetics, clinical evaluation and genetic counselling are delivered predominantly by physicians, with few exceptions (for instance, nurses counsel patients with inherited cancer in some reference centres; genetic information regarding sickle cell trait after newborn screening is given in primary care settings). Genetic counselling is not a recognized health profession.

China**:** Genetic counselling is not a recognized and registered health profession. Genetic counselling is normally carried out by various medical specialties, such as obstetricians, gynaecologists, and paediatricians.

Egypt**:** Genetic counselling services are provided by physicians and genetic counselling is not a recognized health profession.

India**:** Genetic counsellors are not recognised and registered as a health profession. However formal postgraduate training in genetic counselling has been initiated in four institutions. Genetic counselling is provided by medical geneticists as well as other health care professionals.

Oman**:** Medical genetic counselling is not yet officially recognised as a profession. However a PhD programme in genetic counselling is being planned and master degree students in genetic counselling are currently training in the UK.
Genetic counselling is provided by clinical geneticists and paediatricians. Other health care professionals who provide genetic counselling services for haemoglobin disorders within the community genetic programme include medical and nursing staff from primary and secondary health care.

The Philippines**:** Genetic counselling is currently primarily a responsibility of the clinical geneticists. A MSc in genetic counselling programme was started in 2011-2012 at UP-PGH, presently with eight students in the first batch, and six in the second. This MSc in genetic counselling at the UP-PGH caters for nurses, doctors and other allied health professionals. The MSc will train personnel to support services provided by a scarcity of clinical geneticists. The objective is to provide 1 genetic

counsellor in each of the 81 provinces of the country. Medical genetics is being taught by clinical geneticists and visiting genetic counsellors from abroad and includes lectures on the conditions included in the newborn screening panel of disorders.

***South Africa*:** Genetic counselling is a recognised and registered health profession with formal postgraduate training at the master's degree level (requiring 2 years of fulltime formal teaching and clinical training, a research project, and a 2-year internship) and registration with the HPCSA. There are 16 registered genetic counsellors and another four who will become available at the end of 2013. However there are five posts currently available in the country and should any of those posts be vacated they will be frozen and lost.

Genetic counsellors as registered profession

South Africa is the only GenTEE country currently that has non-MD genetic counsellors, educated and trained following the model developed in the UK and in the USA, which is now increasingly being implemented in Europe and elsewhere.

Oman and ***the Philippines*** have initiated training programmes for non-MD genetic counsellors.

Not having genetic counsellors as part of medical genetic services places an increased workload and severe strain on the limited number of physicians available in low- and middle-income countries. Genetic counsellors can alleviate a considerable portion of this workload.

However, genetic counsellors are only part of the solution to the problem. Countries need to train staff in the primary health care (PHC) setting to be able to extend genetic counselling from the tertiary environment into PHC. In ***South Africa***, clinical geneticists suggested training nurses and this worked well. Due to lack of commitment and financial support this programme has become redundant

Education programmes in medical genetics and genetic counselling available for non-genetic health professionals

In all GenTEE countries there are education programmes in medical genetics available for non-genetic health professionals. However, initiatives to provide such training courses depend heavily on the engagement of clinical and other geneticists. In countries such as ***China, India*** and ***South Africa*** governmental support for educational programmes is provided. However, in ***South Africa*** this is very limited and considered to be inadequate to achieve its purpose.

Argentina**:** There are education programmes in medical genetics and genetic counselling available for non-genetic health professionals. Short courses directed at primary care health professionals and general physicians are conducted in urban and rural settings.

Brazil**:** The *Sociedade Brasileira de Genética* (Brazilian Society of Genetics, SBG) has been conducting certifications in human cytogenetics and in human molecular genetics since 1999, through an agreement with the Federal Councils of Biology, Biomedicine and Pharmacy.

China**:** There are several nationwide conferences held by the Central MoH or by the *China Medical Association* (CMA) on the topic of medical genetics and perinatal health care. Major hospitals and institutions also provide short term professional training for non-genetic health professionals.

Egypt**:** Training courses on the detection of congenital and genetic disorders and referral to the community genetic clinics are available for nurses and physicians, in cooperation between the Ain Shams University Department of Paediatrics and the NRC. Physicians working in the community genetic clinics receive condensed practical training courses of two months and can attend specialized courses, for example dysmorphology, premarital counselling, genetic laboratory results interpretation, and prenatal testing and diagnosis.

In ***India***, courses and CMEs are organized by various professional groups. Funding agencies such as the ICMR, the *Council of Scientific and Industrial Research* (CSIR), the government of ***India*** Department of Science and Technology (DST) and Department of Biotechnology (DBT) have task forces in genetics and provide liberal support for educational activities in genetics.

Oman**:** Professional training in genetic counselling for nurses is organised by consultants in clinical genetics for nursing staff. Training seminars in genetic counselling for haemoglobin disorders are offered to primary health care (PHC), maternal and child care workers and physicians within the *National Programme for the Control of Genetic Blood Disorders*.

***The Philippines*:** As mentioned in the previous section, the MSc in genetic counselling programme provides CME on genetic counselling to nurses, doctors and other allied health professionals (such as biologists, behavioural scientists). Health professionals from non-genetic fields are accepted for postgraduate training in molecular biology and biotechnology in UP Diliman, and in molecular medicine in SLMC. The MD PhD programme of the UP College of Medicine offers a PhD in molecular medicine.

***South Africa*:** The National DoH provides very limited finances for medical genetics education for nurses and doctors working in PHC. In 2005 a standardised syllabus for this programme was developed by medical genetics professionals. The programme consists of a four month distance learning section (using a specially compiled manual on birth defects[97]), four contact days of lectures and tutorials, and an examination. Candidates who pass the examination can then undertake four more training days on developing clinical skills, including dysmorphic examination, and counselling skills. Medical geneticists and genetic counsellors give occasional lectures to non-genetic health professions students and qualified health professionals, such as medical specialists and registrars in various medical fields, general practitioners, physiotherapists, occupational therapists, speech therapists, social workers, pharmacists, and nurses, as well as medical insurance personnel.

Education programmes in medical genetics for non-genetic health professionals

The clinical and counselling workload of physicians in medical genetic services in middle-and low-income countries, in addition to being assisted by genetic counsellors, can be further alleviated by training physicians in other disciplines and PHC workers.

It appears that the need for education of non-geneticists in genetics has been recognized widely, but it is also apparent that there is no uniform pattern of programme structures, likely due to variable needs in all GenTEE countries.

[97] Birth Defects: Counselling and caring for children with birth defects. Developed by the Perinatal Education Programme in collaboration with the South African Department of Health, SAIDA and the March of Dimes, EBW Healthcare, 2010. Online available at http://scribd.com/doc/31657154/Birth-Defects-Free-Online-Edition (accessed April 22, 2013)

Brain drain/migration of health care personnel working in medical genetics

Argentina*:** There is some brain drain/migration from ***Argentina to developed countries of health care personnel working in medical genetics but the magnitude is not known. The causes are the same as for brain drain of health care personnel in general, namely low pay, lack of opportunities for advancement, and poor quality of life.

Brazil*:** External brain drain has not been a problem in ***Brazil. Many experts go abroad, especially to the USA and Europe for training, but usually for a limited time. When returning to the country, often new technology is implemented and many services are "upgraded". The absence of medical genetics as a formal medical specialty in the SUS, however, has been leading to a different modality of internal "brain drain", where trained geneticists have no job positions available. Such fact leads trained specialists to other practices / medical specialties where they can earn a living, and many never return to medical genetics again.

China*:** The brain drain/migration of health care personnel working in all fields of medical services is a big problem for ***China, as foreigners with a medical degree awarded by overseas university are not accepted by the National MoH and therefore are not allowed to practice medicine. A number of Chinese-American medical geneticists, who previously studied and were trained in the USA, are involved in teaching and research, but rarely get into clinical practice.

Egypt*:** Brain drain or migration abroad, either temporary or permanent, of medical geneticists is not a significant problem in ***Egypt. Only a small proportion of former staff work outside ***Egypt***, typically in Gulf States where salaries are much higher compared to the small salaries physicians receive in ***Egypt***. The main problem in ***Egypt*** is the inequity in the distribution of genetic specialists in the country, because internal migration to semi-rural, rural and remote areas is not attractive to the majority of physicians with an interest in genetics.

***India*:** PhD students from institutions recognised internationally on graduation often migrate to the West. However it is not possible to quantify the number.

***Oman*:** The brain drain/migration of mostly non-Omani[98] health care personnel working in medical genetics is a problem in the Sultanate of Oman. Around 30% of resignations are due to finding better opportunities elsewhere, the advantage of doing so probably being financial. This impacts negatively on medical genetic health services as it results in insufficient staff numbers.

[98] In Oman, the majority of the heatlh care workfocr comprises expatriates. However, this is slowly changing due to the „Omanization Policy" of the Omani government. Omanization means that skilled occupations hold by expatriates should be filled by trained Omani nationals. Still, many Omani physicians and lab personnel have obtained their training abroad and not in Oman.

The Philippines are strongly affected by brain drain. The majority of the original staff of the IHG-NIH have been absorbed by genetic laboratories overseas, and Philippine students going for PhD work overseas do not return.

South Africa**:** The brain drain/migration of health care personnel working in medical genetics is a problem in the country. Four out of the nine medical geneticists trained in the country between 2000 and 2008 have left the country or the profession. A further three are working part-time and no posts are available at present for future graduates. Around 15% of genetic counsellors have emigrated. Several counsellors are working in part-time positions, including the three who are working in the western Cape. Staffing National Health Laboratory Service (NHLS) laboratories in the public sector is problematic since medical scientists, once qualified, may leave to the private sector for better wages or emigrate. Vacated posts are frozen due to cost constraints. In this way the Division of Human Genetics, NHLS and University of the Witwatersrand (WITS) in Johannesburg, between 2007 and 2010 lost 30% of its laboratory staff. Thus, the country is very short of trained personnel, staff numbers in the field are decreasing instead of increasing, and development in the field is being retarded. Medical scientists in research also leave to gain experience overseas and many do not return.

Brain drain/migration

External (migration) or internal (leaving the field of medical genetics altogether) brain drain due to unsatisfactory career and salary conditions is a problem in all GenTEE countries

Import of specialists from abroad to provide medical genetic services

Import of foreign genetic specialists is not an issue in GenTEE countries except in ***Oman*** where foreign genetic specialists are acting as temporary consultants. This is either because, as in ***Egypt***, enough genetic specialists are available or, as in ***China*** and ***South Africa***, there are difficulties in obtaining registration and low salaries are a deterrent.

Workload

Availability of service and extent of integration of genetic services targeted and designated for public health care into the health care system

In ***Argentina*****,** there are 41 clinical genetic units in the public sector, as part of a national network of 1,319 public hospitals (with a total of 76,885 beds) and 6,290 PHC centres. Some hospital genetic services perform outreach to health centres within their area of influence, but all too often patients affected with genetic conditions must find their own way to a tertiary hospital for genetic services. Some of the few comprehensive genetic centres in tertiary hospitals run telemedicine programmes for genetic consultations accessible to secondary and primary care hospitals in the provinces.

Brazil**:** The Brazilian National MoH published a decree in 2009, which proposes the creation of a *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS).[99] The process that led to acknowledge the need to establish such a policy began in 2001. Some of the conclusions highlighted that most Brazilian regions were hardly prepared for the clinical genetics practice. Thus, with basic problems of infrastructure and shortcomings in the area, the challenge is to establish a minimum organizational structure, from which strategic actions would be applied to ensure comprehensive care in genetics.

China**:** Clinical genetic services, although limited, are integrated into the public health system, in and around the academic centres of the universities and hospitals. Most genetic counselling clinics are held in public hospitals, especially maternal & child health hospitals. The medical genetic staff hardly undertake outreach visits due to the limited resources, the majority of the population still remain underserviced. An efficient hierarchical maternal & child health care network has been established from small towns to metropolitan cities. The right of frequent medical examination/screening during pregnancy is protected by the *Law of the People's Republic of China on Maternal and Infant Health Care* (MIHCL)[100] enacted in 1995. Facilities for general prenatal screening, karyotyping, biochemical tests, ultrasonography screening, microbiological tests are present in all level hospitals; PND is only available in qualified centres in major cities.

Egypt**:** Genetic services are integrated into the primary, secondary and tertiary health care. The community genetic counselling clinics are a referral site between primary and tertiary care. The clinics work through a system of referrals from primary

[99] Brasil (2009a), Ministério da Saúde, portaria GM no. 81, 20 de janeiro de 2009 – Institui, no ambito do Sistema Único de Saude (SUS), a Politica Nacional de Atenção Integral em Genetica Clinica. Diário Oficial da União 21/01/2009

[100] The Law of the People's Republic of China on Maternal and Infant Health Care. Online available at www.asianlii.org (accessed May 13, 2013)

care to secondary care then tertiary care level of services. The early detection of genetic and congenital disorders is done at PHC level, and then cases are referred to the genetic counselling clinics in the catchment area where diagnostic services, counselling and follow-up are provided by secondary care physicians. Investigations, treatment and rehabilitation services are provided by the health insurance organization, and then cases are referred again to the clinics for follow-up.
The general population has also a direct access to a tertiary level of medical genetic care through the genetic departments, centres, and units at university level and the NRC. Nevertheless, at that level there are no service networking activities between the different universities.

***India*:** Most genetic centres are part of hospitals providing comprehensive care. Many of the hospitals are in the government sector, and thus represent tertiary care referral centres for primary and secondary health care. There are some laboratories which are quaternary centres, national laboratories that obtain specimens from throughout the country, and not linked with hospitals.

***Oman*:** Services for haemoglobin disorders have been integrated into the PHC system.

***The Philippines*:** In 2004 newborn screening was integrated into the public health delivery system. All newborn screening laboratories are considered public health laboratories. Guidelines and accreditation are run by the DoH. Aside from newborn screening, the clinical genetics unit of the department of pediatrics, PGH, receives consultations and referrals for genetics-related cases from all over the country. The *Telegenetics Referral System* (TRS) is undergoing pilot implementation in 10 hospitals all over the country, with the aim of providing genetic services in remote areas of the country.

***South Africa*:** Clinical genetic services, although limited, are integrated into the public health system, in and around the academic centres of the universities of Cape Town, Free State, Stellenbosch, WITS and formerly KwaZulu Natal. Most genetic counselling clinics are held in public hospitals, associated with academic hospitals and the genetic services are available through referral from other hospitals and clinics. The medical geneticists and genetic counsellors also undertake outreach visits both within and outside their provinces, although, with the limited resources, the majority of the population still remain underserved.
Community education of health care professionals, at many levels from PHC to tertiary settings, is also undertaken in order to increase awareness and integrate the services into the healthcare system. At present, a limited number of nurses in PHC are receiving basic in-service training in medical genetics, and paramedical professionals, such as physiotherapists and occupational therapists, receive some teaching during their training.

Extent of integration of genetic services into the public health care system

Clinical genetic services are integrated – although to a limited extent – into the public health care system in the GenTEE countries. Most of these services are available at secondary or tertiary care level in urban areas. In the rural areas, the availability of genetic services is still scarce.

While genetic services appear to be integrated into the health care system, the majority of genetic patients are clearly underserviced in all GenTEE countries.

Number, location and regional distribution of medical genetic departments/medical genetic units/centres

Table 6.2 Number of "genetic units"[101] and laboratories in GenTEE countries (2010)

country	genetic units	laboratories/integrated laboratories
Argentina	41[102] (public domain)	29 public 3 private
Brazil	66	47 public 50 private, of these 25% are exclusively genetic laboratories
China	- 9 university bound medical genetic centres - 2 reproductive centres (Jiangsu, Yunan) - unknown number of PND centres at major provincial hospitals	75 public number of private laboratories unknown (mainly in the Beijing, Shanghai and Guangzhou area)
Egypt	11 genetic counselling clinics 5 genetic departments in university hospitals & a genetic division with 8 departments at the NRC	8 public 10 private
India	54[103]	10 public 35 private
Oman[104]	2 MoH services 5 SQU	3 public (2 MoH 1 forensic) 5 university bound (Ministry of Education)
Philippines	1 (Clinical genetics Unit, Department of Pediatrics, PGH)	4 Newborn screening (3 public, 1 private) 3 Molecular (1 public, 2 private) 1 Biochemical (1 public) 4 Cytogenetics (2 public, 2 private) 4 DNA analysis (paternity testing, 3 public, 1 private)
South Africa	4[105]	8 public 5 private

[101] Definition of "genetic unit": A genetic unit is a clinical entity staffed by clinical geneticists and provides access to laboratory diagnoses.
[102] Of these only 5 genetic units conform with the notion of "comprehensive genetic centre".
[103] Cytogenetic investigations are being undertaken by 45 centers, biochemical investigations at 28 centers, PND by 45 centers, and genetic counselling at 45 centers. Delhi has 6 centers, each of which serves 2.3 million people.
[104] Genetic service units and laboratories in the public domain.
[105] Division of Human Genetics laboratory at the NHLS and the University of the Witswatersrand, Johannesburg, University of Cape Town, Division of Human Genetic laboratory and Groote Schuur Hospital, University of Stellenbosch/Tygerberg Hospital laboratory, University of the Free State, Human Genetic laboratory

Location and regional distribution of genetic services

In all GenTEE countries considering their populations, there are not sufficient medical genetic units available.

For instance only 54 genetic units are presently running in different parts of ***India***. This is considered insufficient for a large country like ***India***, **where some capitals of the 28 states do not have genetic services**.

Genetic units are nearly always based in tertiary care at university hospitals and thus are likely to serve the more affluent urban middle and upper classes and to a lesser extent the rural population.

In ***the Philippines*** only PGH (the largest tertiary government hospital in the country) has a clinical genetics unit. Most of the geneticists in the country are located in the Manila area, particularly in PGH. There are efforts to change this now, through the TRS. However, this system does not involve setting up more genetic units. Laboratories are also mainly at UP Manila. Samples from all over the country are received here. There are private laboratories but their services cannot be afforded by the less affluent population.

Access to genetic testing and most common referrals for genetic testing

In all GenTEE countries except ***Oman*** there is a mix of private and public laboratory service distributed over the country, but with a focus on big cities and poorer access in rural areas. The laboratories offer a broad range of genetic laboratory techniques to cover the needs for clinical diagnosis.

In ***Brazil***, ***China***, ***India*** and ***Oman*** all established laboratory techniques and NGS technology are available to support diagnostic services within the country (**Table 6.3**).

Table 6.3 Availability of different genetic testing techniques in GenTEE countries (2012)

Technique	Argentina	Brazil	China	Egypt	India[106] (2010)	Oman	Philippines	South Africa[107] (2007)
Conventional Cytogenetic Techniques constitutional	●	●	●	●	●	●	●	●
Conventional Cytogenetic Techniques acquired	●	●	●	●	●	●		●
mFISH	●	●	●	●	●	●		●
iFISH	●	●	●	●	●	●		●
MLPA		●	●		●	●		●
PCR/ sequencing	●	●	●	●	●	●	●	●
QF-PCR	●	●	●		●	●		●
RT-PCR	●	●	●	●	●	●	●	●
Southern Blotting	●	●	●	●	●	●		●
Microarray		●	●		●	●[108]		
Metabolic Biochemistry	●	●	●	●	●	●[109]	●	●
NGS (Next Generation Sequencing)		●	●		●	●[110]		

● = available

mFiSH Multicolor fluorescence in situ hybridization
iFiSH Interface fluorescence in situ hybridization
MLPA Multiplex ligation-dependent probe amplification
PCR Polymerase chain reaction
QF-PCR Quantitative fluorescent polymerase chain reaction
RT-PCR Reverse transcription polymerase chain reaction

[106] Sir Ganga Ram Hospital, Center of Medical Genetics: Directory of Genetic Tests, 2010. Available from: http://geneticsindia.org/FullTestList.aspx?lab_id=1 (accessed May 16, 2013)
[107] Department of Health, South Africa (2007). Diagnostic Genetic Tests. Available from: www.doh.gov.za/docs/index.html (accessed May 16, 2013)
[108] At Sultan Qaboos University
[109] Available at university hospital
[110] From 2012

The most common referral for a genetic investigation is dysmorphology in all countries except ***Oman*** (**Table 6.4**).This is probably a reflection of the inherited disease pattern of the country; haemoglobin disorders being the most common inherited disorder in the country. Compared to the other GenTEE countries, ***Oman*** has a slightly different pattern for referring to genetic testing also reflecting the structure of the genetic burden in the society.

Table 6.4 Ranking (1-10) among GenTEE countries of the ten most common indicators for issuing a genetic test (estimates by the GenTEE consortium)

Example for common indicators	Argentina	Brazil	China	Egypt	India	Oman*	Philippines	South Africa
Nonsyndromic Mental Retardation	3	2	3	3	1	5	2	2
Neuro-Muscular Disorders	2	6	4	8	5		4	3
Haemoglobin disorders		9	7	6	2	1	5	8
Congenital Malformations/Dysmorphic	1	1	1	1	3	2	1	1
Cancers (familial)	7	4	9	9	9	3[111]	7	5
Infertility, including recurrent miscarriage	4	8	5	4	4	4	6	7
Profound Deafness (Childhood)	8	7	6	7	7		9	10
Family History, including Premarital Counselling, excluding cancers and CF	5	5	2	5	6		8	6
CF	9	10	10	10	8	10	10	4
Failure to Thrive	6	3	8	2			3	9

*Additional ranking of disorders not included in the table for Oman:
Repeated Stillbirth 6
Growth failure in a child, delayed puberty or ambiguous genitalia 7
Chromosomal fragility testing 8
Premature ovarian failure 9
Multiple affected birth with spinal muscular atrophy, Osteopetrosis, cystic fibrosis 10

[111] Hematological cancer

Quality assurance of medical genetic services

Availability of quality assessment schemes and existing regulatory frameworks for genetic services

In GenTEE countries the healthcare systems are multifaceted and **there is neither official framework for assessing new genetic tests that become available nor any formal system for approving which tests may be used in a clinical setting. Accreditation of clinical laboratories is not mandatory in most of the countries**.

Clinical laboratories are accredited by special agencies of the ministries of health in ***Argentina***. Except for laboratories that perform newborn screening, there are no official agencies that control or monitor the analytical validity of tests. **Quality assessment of laboratory results relies mostly on the voluntary decision of the laboratories directors to participate in a quality control programme, usually of an international agency.** Chromosome and DNA studies are performed in the laboratories of public hospitals, mostly teaching hospitals, or by private laboratories. While these laboratories are certified by a state agency, **participation in quality assessment programmes is voluntary and regulation very lax**. Further, while in theory most of these laboratories must have in-house and external quality control programmes, there is little government oversight on these issues.

In ***Brazil***, there is **no specific regulation for medical genetics services**. All other medical services are regulated and supervised by the National MoH and its specific agencies, particularly the *Agência de Vigilância Sanitária* (ANVISA)[112]. Some quality assessment programmes are available for laboratories, so that they comply with international standards. **Most private laboratories tend to undertake it voluntarily, not specifically for genetics, but for all testing offered.**

In ***China***, quality assessment schemes are available and the centres in hospitals are exposed to regular peer reviews. **All clinical laboratories offering genetic diagnosis are required to meet the standards of the Central MoH's *Centre of Clinical Testing*. Some private laboratories may also comply with ISO15189 (Accreditation Criteria for the Quality and Competence of Medical Laboratories) and obtain accreditation from *College of American Pathologists* (CAP).**

There are quality assessment schemes for genetic laboratories in ***Egypt***, either internal from within the laboratory itself or external, through a protocol of agreement with an external laboratory from another country, e.g. the CDC quality assurance for the MoH&P central health laboratories, especially for neonatal screening tests.

[112] ANVISA, 2010. Agencia Nacional de Vigilância Sanitária http://portal.anvisa.gov.br/wps/portal/anvisa/home/servicosdesaude (accessed August 29, 2010.

In **India** the *National Accreditation Board for Testing and Calibration of Laboratories* (NABL) inspects and accredits the laboratories for genetic tests. The quality assurance programme involving exchange of samples is not done by this board. A quality assurance programme for thalassaemia and haemophilia is run by the Department of Haematology Christian Medical College, Vellore. Some laboratories also enrol in the *European Molecular Genetics Quality Network* (EMQN) programme of the EU for molecular tests. Many centres performing newborn screening tests are enrolled in the quality control programme run by CDC.

The human genetic laboratories of the MoH in ***Oman*** follow the quality assurance for cytogenetic testing of blood, bone marrow and amniotic fluid available from the *Cytogenetic European Quality Assessment* (CEQA) [113] since 2009, but **quality assessment schemes are not available locally.**

The Philippines follow both internal and external quality assessment schemes, like the CEQA. The newborn screening laboratories have been participating in the proficiency testing of the CDC since 1997 while the cytogenetics laboratory started only last 2011.

In ***South Africa***, some quality assessment schemes are available and the academic departments of human genetics are exposed to regular peer reviews. NHLS laboratories in academic and tertiary settings, doing genetic testing, can be monitored and accredited by the *South African National Accreditation System* (SANAS) so that they comply with international standards. However, this process is not mandatory. **Private laboratories are not subjected to such scrutiny but tend to undertake it voluntarily.** The NHLS laboratory at WITS University participates in biannual quality assurance programmes and obtains accreditation for testing proficiency through the CAP. It is in the process of moving into new accommodation and when this is complete it will undertake SANAS accreditation. Further, biological medical scientists follow a set syllabus and intern programme laid out by the HPCSA and only HPCSA registered medical scientists may work in the public and private sector laboratories.

Due to the variability of health care systems in the GenTEE countries it appears that the flexible European quality assurance systems could serve as an optimal model for emerging economies.

[113] http://www.eurogentest.org (accessed May 16, 2013)

Harmonization of quality assurance through international networking

In Europe, quality assurance within genetic testing laboratories is guided largely by a network of external quality assurance (EQA) providers, which provide laboratories with genetic test samples and feedback based on the laboratory's analysis of the sample, both in the form of individual reports and group feedback that can lead to standardized guidelines. These providers vary in the number of tests schemes (programmes that test laboratory proficiency by examining testing results on a standardized sample) available and geographical scope The EMQN and the Cystic Fibrosis Thematic Network are among the largest providers, and both have received funding from the EU. Additionally, it may be beneficial in the longer term for other jurisdictions, such as the Latin American countries, and the other GenTEE countries to participate in similar harmonization activities, as they may benefit more from this model than from one developed for a single country (e.g. the United States), because Latin American countries face similar challenges regarding the sufficient availability of testing samples. It may be possible as well to preliminarily include developing countries to join in under the umbrella of a European cross-national quality assurance plan. Indeed, the EMQN welcomes countries outside Europe within it schemes. Given multiple regional or continental networks of quality assurance, international groupings can themselves interact to produce a truly worldwide harmonization of quality.

Documentation of process and outcome data

The documentation and process of outcome data, takes place frequently at the level of each institution providing the service and no national data are available, like in ***Argentina*** and ***Brazil***, ***China***, ***Egypt***, ***South Africa***.

In ***Argentina*****,** the documentation of process and outcome data on medical genetics services takes place at the level of each institution providing the service and no national data are available. Each medical centre keeps the records and data they require for their internal assessments and/or annual reports and there is no national policy or coordination of data for the whole country.

Brazil has a good quality and data control in the newborn screening programme. All newborn screening centres have to undergo an initial accreditation and are only promoted to the more complex phases (more diseases screened) based on quality standards for uptake and results. The National MoH has a permanent commission for the follow up of this programme.

In ***China***, only the centres in hospitals are required to submit annual report to Maternal & Social Health Section, Central MoH, but national data are not accessible for the public. Each academic centre keeps the records and data in their own database, and there is no policy to update the records to administration units.

In ***India***, accreditation by the NABL involves lot of documents that are to be submitted to the board before the physical inspection. Most genetic centres maintain registers of the patients with genetic disorders seen by them.

In ***Egypt***, each genetic laboratory or genetic department or centre (either academic or private) has its own records and annual data that is not shared on a national level and the MoH&P does not have a direct authority to have access to those records.

In ***Oman***, yearly reports to the MoH have to be prepared on: clinical consultations, laboratory performance, quality measures and training activities.

In ***the Philippines***, all newborn screening centres undergo an initial accreditation and re-accreditation every three years. The accreditation team consists of both local and international experts.

For ***South Africa***, each academic centre keeps the records and data they require for their internal assessments and/or annual report and there is no national policy or coordination of data for the whole country.

Availability of national guidelines and recommendations for the provision of medical genetic services including ethical guidelines

National guidelines or recommendations for the provision of medical genetic services are rare in GenTEE countries. ***Argentina*** and ***Egypt*** have no national guidelines, ***the Philippines*** have guidelines for the provision of newborn screening.[114]

In ***Brazil***, aiming to assist in medical decision making and thus optimize the care of patients, the AMB and the CFM in 1999 triggered a process with the specialty societies for the development of medical guidelines based on scientific evidence currently available. Since 2000, the SBGM contributed seven guidelines: *(i) Clinical Genetics Evaluation of the Newborn, (ii) Familial Cancer, (iii) Female Sterilization: Statement, (iv) Male Sterilization: Indications, (v) Laboratory Tests for Diagnosis of Symptomatic Diseases, (vi) Turner Syndrome: Diagnosis and Treatment, and (vii) Predictive Testing (CFM projeto diretrizes)*. In 2010, the SBGM started working intensively in elaborating more than 40 guidelines, especially in regard to diagnostic tests and new treatments[115]. Official government documents are rare, but in the last decade some important initiatives are being taken. In 2001, the National MoH established a commission to discuss and propose recommendations related with the access and use of the human genome. This resulted in a document published in

[114] Newborn Screening Act of 2004 (RA 9288)
[115] data obtained by Dafne Horovitz by personal communication

2003[116] with recommendations about genetic tests and the ethical use of genetic information. The document recommends that:

- diagnostic genetic tests, predictive or not, performed in the context of research involving humans, are voluntary, after proper guidance, and always preceded by the signing of informed consent (except tests of public health programmes, as neonatal screening, and those that aim to reduce personal risk to health or health of third parties);

- tests that are performed with a medical purpose in order to diagnose and establish appropriate schemes of therapy and prevention should not be indicated before its sensitivity, specificity and efficacy have been scientifically substantiated;

- it is forbidden to request genetic tests as a prerequisite for job admission, except as provided in specific legislation, or granting benefits, by any public or private institution, or that the personal genetic information is used in a discriminatory manner in such institutions;

- it is prohibited to disclose genetic test results to any person other than the individual himself/herself or his/her legal representative;

- genetic testing, both diagnostic and predictive, can only be performed on medical request and

- that education for health professionals and for the population is highly recommended, in order to clarify the benefits and risks of information obtained from genetic tests.

In *China*, the *MIHCL* provides recommendations for the provision of genetic services. In 2001, the ***China*** State Council published a *State Council Order (No. 308)* on the implementation of the *MIHCL*. The law and the council order provide a detailed guideline for the development of maternal & child health care, especially in the area related to the "detection and control" of genetic disorders.[117]

The Central MoH has published a list of regulations, these include:

[116] CAGH (2003) - Comissão sobre Acesso e uso do Genoma Humano- Informação genética- Testes genéticos e recomendações (2003). Ministério da Saúde, Secretaria de Ciência, Tecnologia e Insumos Estratégicos. Departamento de Ciência e Tecnologia. Online available at http://www.ghente.org/temas/informacao/testes_geneticos.pdf (accessed May 6, 2013)

[117] For the controversy surrounding the MIHCL and its subsequent development see:
Hesketh T, Wei XZ: Maternal and child health in China. BMJ 1997;314:1898. Online available at http://www.bmj.com/content/314/7098/1898?view=long&pmid=9224139 (accessed May 13, 2013)
Guo SW: China: The Maternal and Infant Health Care Law, Fundan University Shanghai, China. Abstract available online at http://onlinelibrary.wiley.com/doi/10.1002/9780470015902.a0005201.pub2/abstract (accessed May 13, 2013)

- implementation of human Assisted Reproductive Technology (ART);
- management of human ART;
- specifications for human ART;
- principle consideration of ethical issues in ART;
- basic standards for human sperm bank;
- sperm bank management practices;
- technical specifications of sperm bank.

The right for people to access genetic services is legally protected, professional regulations are monitored by the Central MoH and services (including newborn screening, cytogenetic testing and PND) have been established in most regions (see **Figure 4.3** above) at provincial level. All the service centres (including both university and hospital-based ones) are registered by the Central MoH. The quality of services is monitored once every two years.

In ***India***, the government has become very strict and has mandated that all institutions carrying out research should have an ethics review board.
The ICMR has issued *Ethical Guidelines for Biomedical Research on Human Participants* in 2006[118]. 13 pages cover ethical guidelines for genetic research. These include: general guidelines, pedigree studies, participant requirements, informed consent, and confidentiality of data and defines risk and benefits. There is a separate section on genetic screening that includes prenatal testing, screening in newborns, screening of children and anonymous testing. Another section covers therapeutic trials including gene therapy, the human genome project, DNA and cell banking/repository and DNA diagnosis and PND. There are special regulatory bodies that are tasked to ensure the review, approval and monitoring of all research projects in the field of stem cell research.[119]

In ***Oman***, the MoH adopted the WHO *"Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetic Services"*[120] in 1998, omitting the topics of MToP and PND.

In ***South Africa***, *National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities*, were initially drawn up with contributions from all major stakeholders (from academic and government departments) and a document was published in 2001 by the National DoH[121]. The document provides recommendations for the provision of genetic services and has

[118] http://icmr.nic.in/ethical_guidelines.pdf (accessed May 7, 2013)
[119] http://icmr.nic.in/stem_cell_guidelines.pdf (accessed April 11, 2013)
[120] http://www.who.int/genomics/publications/en/ethicalguidelines1998.pdf (accessed April 11, 2013)
[121] http://www.westerncape.gov.za/text/2003/humangenetics.pdf (accessed May 2, 2013)

sections on general ethical guidelines for medical genetics (modified from the WHO *Hereditary Disease Programme*, 1996, document[122]) and on ethical principles for genetic professionals.[123] This policy document is still available and it has not yet been superseded.

Some ethical guidelines, for genetic research purposes, were drawn up, around the same time, by the *Medical Research Council* (MRC) and a committee was set up for the purpose. These guidelines appeared in a booklet entitled: *Guidelines on Ethics for Medical Research: reproductive biology and genetic research*.[124] This booklet followed on from a booklet on the general principles of ethics for medical research.[125]

Setting international standards

GenTEE countries face a similar situation as the USA and many European countries. These countries, despite their relatively advanced state in developing and coordinating the various institutions involved in setting standards at different levels of the process, have yet to succeed in fully integrating what continues to be a somewhat fragmented structure. The OECD, the EC and the WHO have been working together in recent years to develop consensus around international standards and best practices for ensuring the quality of genetic services in their member countries. There remains much work to do to finalize and implement their recommendations.

A number of bodies have made recommendations relating to quality assurance in healthcare, and specifically in the provision of genetic services. Many calls have been made for the creation of an independent and autonomous national level body assigned the task of institutionalizing a quality assurance process across an entire country. The WHO has also recommended that a national health laboratory policy should be developed, emphasizing quality assurance and the networking of laboratories. Such a policy should be backed by legislation and overseen by a broad based group comprising a range of stakeholders, recognizing that a successful system must take into account the widely differing perspectives and motivations of the stakeholders involved.

[122] WHO Technical Report Series 865: Control of Hereditary Diseases, Geneva, 1996. Online available at http://whqlibdoc.who.int/trs/WHO_TRS_865.pdf (accessed April 22, 2013)
[123] Baumiller RC, Cunningham G, Fisher N, Fox L, Henderson R, Lebel R, McGrath G, Pelias MZ, Porter I, Seydel F, Wilson NR (1996). Code of Ethical Principles for Genetics Professionals. Am J Med Genet; 65: 177-183.
[124] Medical Research Council, South Africa (2002). Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research. MRC, South Africa, Tygerberg.
[125] Medical Research Council, South Africa (2002). Guidelines on Ethics for Medical Research: General Principles. MRC South Africa, Tygerberg

National policies and legal frameworks

Existing national policies, guidelines and planning activities for the provision of medical genetic services

In most GenTEE countries, the development of national policies, guidelines and planning activities for the provision of medical genetic services started in the 2000s, reflecting the advances in genetic technologies and genetic sciences development.

Argentina*:** There are no national guidelines or recommendations for the provision of medical genetic services, including ethical guidelines. The national policies and legal frameworks regarding medical genetics are within the responsibility of the National MoH. In turn, the National MoH has been influenced by experts in medical genetics from the Pan American Health Organization (PAHO). Since 1982, PAHO has sporadically been convening regional consultations of experts in medical genetics, which in turn issued recommendations to member countries, including ***Argentina.[126,127 ,128,129,130,131] A special consultation took place in ***Argentina*** in 2003, in which specific recommendations were issued by the consultant, including the appointment of a blue ribbon *National Commission on Genetics and Health* that would have the task of surveying the status of medical genetics services in the country and pointing needs for development.[132] This National Commission was created in 2006 and has since conducted a countrywide survey of genetic services and formulated a plan for their strengthening and coordination.[133] Currently, the National MoH has adopted as a national policy this report of the National Commission and has been strengthening the network of genetic services in the public sector as well as developing a nationwide registry of congenital anomalies, which has already started in the northeastern provinces of the country.[134]

[126] Pan American Health Organization (1984). Prevention and Control of Genetic Diseases and Congenital Defects. Report of an Advisory Group (Castilla EE, Penchaszadeh VB, Wertelecki W, Youlton R). Scientif Publ. 460. Washington, DC, PAHO, 1984.
[127] Penchaszadeh VB and Beiguelman B (1998): Medical genetics services in Latin America: Report of a meeting of experts. Pan Am J Public Health 3(6): 409-420, 1998.
[128] Kofman-Alfaro S, Penchaszadeh VB (2004). Community Genetic Services in Latin America and Regional Network of Genetic Services. Community Genet 7:157-159, 2004.
[129] Penchaszadeh VB (Editor) (2004). Medical Genetics in Latin America. Community Genet 7: 2-3.
[130] Pan American Health Organization (2007a). Program on genetics, public health and human rights. Unpublished document. Available from Victor Penchaszadeh.
[131] Pan American Health Organization (2007b). Recommendations for the development of genetics in public health in Latin America. Report of a meeting of experts. Rio de Janeiro, June 21, 2007. Unpublished document. Available from Victor Penchaszadeh.
[132] Penchaszadeh VB (2003): Medical Genetics in Argentina: Assessment and perspectives. Report of a Consultation to the Pan American Health Organization for the Ministry of Health of Argentina. Unpublished document, available from the author.
[133] Alba A, Barbero P, Barreiro C, Chertkoff L, Dain L, Ferreiro V, Francipane L, Frechtel G, Gallego M, Liascovich R, Meroni ME, Rozental S (2007). Diseño y organización de una Red Nacional de Genética Médica (Design and organization of a national network of medical genetics.) Spanish. Unpublished, available from Victor Penchaszadeh.
[134] Liascovich R, Gili J, Valdez R, Somaruga L et al (2011). Development of a national registry of congenital anomalies in Argentine: a pilot feasibility study. Rev Argent Salud Publica 2 (6):6-11.

***Brazil*:** The Brazilian National MoH published a decree in 2009, which proposes the creation of a *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS).[135]

The process that led to acknowledge the need to establish such a policy began in 2001 and was partly influenced by the announcement of the sequencing of the human genome. The ethical, political, legal, and administrative matters related to the access to human genetic material became an issue in most countries, and the national *Comissão sobre Acesso e Uso do Genoma Humano* (Committee on Access and Use of the Human Genome) was created[136].

During the period 2004–2006, there were several meetings and two regional workshops (south/ southeast and north/northeast/center-west). Representatives of the National MoH also participated in inserts during clinical genetic conferences.
This process resulted in a proposal that ultimately led to the ordinance No. 81 of the National MoH in January 20th 2009, which established the *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS)[137], and also designated the strategies for actions that must be taken into account in its regulation.

The existence of a published policy for genetics constitutes an important historical milestone for ***Brazil***. By the time this text was written, however, no supplementary ordinance, which would be absolutely essential to organize and regulate this policy, had been published. Since January 2009, the *Aliança Brasileira de Genética* (Brazilian Genetic Alliance)[138], several patient–parent organizations and the SBGM have been trying to pressure the National MoH to implement the policy. In 2012 a commission for the elaboration of a policy for rare diseases was constituted, as a result from pressures from these organizations. It is expected that the policy will be instituted in 2013.

***China*:** The *MIHCL* legally guarantees that every Chinese national registered at birth has the right to have access to health care. One part of national policy prioritises services for the prevention of congenital disorders. Guidelines for preconception and pregnancy health care services were approved by the Central MoH in 2011. The guidelines clearly outline the standard of services required prior to conception, during pregnancy, at birth, in infancy and childhood and the way in which these services could be delivered at various levels from primary to tertiary health care settings.

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[135] Brasil (2009), Ministério da Saúde, portaria GM no. 81, 20 de janeiro de 2009 – Institui, no ambito do Sistema Único de Saude (SUS), a Politica Nacional de Atenção Integral em Genetica Clinica. Diário Oficial da União 21/01/2009
[136] Brasil, Ministério da Saúde (2001). Portaria N.º 470/GM, de 6 de abril de 2001 – Institui a comissão sobre acesso e uso do genoma humano.
[137] Brasil (2009), Ministério da Saúde, portaria GM no. 81, 20 de janeiro de 2009 – Institui, no ambito do Sistema Único de Saude (SUS), a Politica Nacional de Atenção Integral em Genetica Clinica. Diário Oficial da União 21/01/2009
[138] http://www.abg.org.br/abg/abg.asp (accessed April 15, 2013)

The use (including for research purposes) of blood products, tissue, stem cells and gametes and zygotes in humans are regulated. Any use or research of these materials requires a written application to and permission from a local ethics committee.

In 2002, the *National Population and Family Planning Commission*, Ministry of Health, and State Food and Drug Administration jointly issued a *"Regulation of the prohibition of foetal sex determination and pregnancy termination for non-medical purposes"*. The regulation defines the conditions and procedures to be adhered to when determining the sex of the foetus and when inducing a sex-selective termination of a pregnancy. Foetal sex determination and sex-selective induced abortion for non-medical purposes are illegal and violations are subject to criminal sanctions and loss of license to practice. Parents and medical professionals are not allowed to use foetal sex for the termination of a pregnancy for reasons other than medical purposes.[139] Termination of pregnancy is prohibited for any reason after 28 weeks of gestation.

In addition to the legislation set out above there is a guideline for the use of informed consent document (issued by the Central MoH) to protect autonomy and privacy.

In Egypt, policies and planning activities related to the provision of genetic services are included under the MoH&P five-year plan for the prevention and early intervention of disabilities:
The MoH&P five-year plan addresses the following dimensions:

- an integrated system for treatment and rehabilitation by implementing registries;

- a prevention and early detection programme by establishing

(i) a national premarital care counselling programme;

(ii) a safe motherhood programme which covers antenatal care, childbirth care, neonatal care and post-natal care (focusing on congenital anomalies, jaundice and early detection of causes of mental retardation)[140];

(iii) a national newborn screening programme for CH[141];

[139] In the People's Republic of China PND is often used to determine foetal sex and is followed by the subsequent abortion of female foetuses. This has led to a severe imbalance of the female/male sex ratio at birth. Despite the regulation the loss of female births due to illegal pregnancy termination for non-medical reasons still seems to be continuing.
[140] Within this domain, a number of screening tests are introduced in the service; however, the focus is still directed towards family planning to ameliorate the serious problem of excessive population growth. Iron-folic acid tablets for treatment of anaemia and prevention of neural tube defects are provided via this programme.

and

(iv)a programme monitoring child growth and development, immunization, nutrition and care;

- rehabilitation through supporting rehabilitation centers and introduction of community-based rehabilitation (CBR).

India*:** There are no national policies guidelines in planning activities for provision of medical genetic services in ***India. Guidelines for research have been issued by the ICMR and theses are enumerated below. There are also guidelines for ART.
As PND was (and still is[142]) often used for the diagnosis of foetal sex for non-medical purposes and abortion of female foetuses the government promulgated a law entitled *"Pre-Natal Diagnostic Techniques (PNDT) Act"*, in 1994. The PNDT Act was implemented in 1996[143] and was amended in 2002 bringing IVF and ART within the framework. The amended bill was entitled *"Pre-Conception and Pre-Natal Diagnostic Techniques (Prohibition of Sex Selection) Act"*. A further amendment in 2003 brought all ultrasound examinations during pregnancy also under its ambit. Every ultrasound machine had to be registered. After numerous complaints from radiologists the range of indications for which ultrasound examinations were permitted were listed.
The ICMR has issued the following guidelines:

- ART (Regulation) Rules[144];
- National Guidelines on the Management of Retinoblastoma[145];
- Guidelines for Good Clinical Laboratory Practices[146];
- Guidelines for Stem Cell Research and Therapy[147];
- Guidelines for Management of Type 2 Diabetes[148];
- Guidelines for International Collaboration/Research Projects in Health Research[149];
- Ethical Guidelines for Biomedical Research on Human Participants[150];
- Intellectual Property Rights Policy[151];

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[141] implemented in 2000, by the end of 2003 all 29 governorates were covered. Pilot testing for expanding the programme to include screening for PKU and Galactosaemia have shown promising results.
[142] Subramanian SV, Selvaraj S: Social analysis of sex imbalance in India: before and after the implementation of the Pre-Natal Diagnostic Techniques (PNDT) Act. J Epidemiol Community Health. 2009; 63:245–252. Online available at http://jech.bmj.com/content/63/3/245.full.pdf (accessed May 13, 2013)
[143] Government of India. Annual report on implementation of the Pre-conception and Pre-Natal Diagnostic Techniques (Prohibition of Sex Selection) Act. New Delhi: PNDT Division, Ministry of Health and Family Welfare, Government of India, 2005. Online available at http://www.pndt.gov.in/writereaddata/mainlinkfile/file22.pdf (accessed May 13, 2013)
[144] http://icmr.nic.in/guide/ART%20REGULATION%20Draft%20Rules%201.pdf (accessed May 8, 2013)
[145] http://icmr.nic.in/guide/RB_Guidelines.pdf (accessed May 8, 2013)
[146] http://icmr.nic.in/guidelines/GCLP.pdf (accessed May 8, 2013)
[147] http://icmr.nic.in/stem_cell_guidelines.pdf (accessed May 8, 2013)
[148] http://icmr.nic.in/guidelines_diabetes/prelim.pdf (accessed May 8, 2013)
[149] http://www.icmr.nic.in/guide.htm (accessed May 8, 2013)
[150] http://icmr.nic.in/ethical_guidelines.pdf (accessed May 8, 2013)

- National Guidelines for Accreditation, Supervision and Regulation of. ART Clinics[152].

Oman: The MoH of The Sultanate of Oman has recognized the need for appropriate medical genetic services and genetic technologies to control congenital/genetic disorders. The *National Committee for the Prevention of Genetic Diseases* was established in 2004 and includes representatives from the Ministries of Health, Education, Social Affairs, Information and National Economy.
In 2005, the MoH published its *7th Five-Year Plan for Health Development* (2006-2010)[153] which included a national strategic plan on genetic diseases in order to reduce the morbidity and mortality.

Its objectives were

(i) improving the management of patients affected by congenital/genetic disorders (especially for congenital blood diseases, mental retardation including Down syndrome and physically impairing conditions), and providing effective preventive measures;

(ii) developing molecular genetic technology expertise capable of supporting local care and prevention programmes;

(iii) providing premarital services to inform about increased risks for congenital/genetic disorders including risk assessment;

(iv) providing public health education on genetic risks for the Omani population.

To achieve its objectives, the National Genetic Centre was established to provide clinical and laboratory diagnostic services, care services and prevention programmes and to conduct training activities and research in the field of medical genetics was enacted. The National Genetic Centre has been endowed with a budget of 5.2 million R.O. (~ 10,288,000 €/13,467,000 US$ in April 2013).

The demand on national manpower development was appreciated and a number of Omani nationals are currently training in the Sultanate and abroad in the field of genetic medicine and genetic laboratory technologies.

[151] http://www.iitr.ac.in/ipr/IPR%20Policy.pdf (accessed May 8, 2013)
[152] http://icmr.nic.in/art/Prilim_Pages.pdf (accessed May 8, 2013)
[153] Ministry of Health, Sultanate of Oman (2005) 7th Five-Year Plan for Health Development (2006-2010). Available at http://www.moh.gov.om/en/nv_menu.php?o=fiveyearplan/fiveyearPlan.htm&SP=1 (accessed 17 October, 2012)

Existing national policies are within a framework of Muslim Law (Sharia) which forbids the interruption of pregnancies after 12 weeks gestation exempt for cases where there is danger to the mother's life or where ultrasound reveals structural anomalies incompatible with life such as hydrocephaly, anencephaly, renal dysplasia. The rising number of surviving disabled children with congenital and genetic disorders is creating awareness and concern about the increase of families with disabled children. The national policy for prevention is to avoid marriages between carriers and in correspondence with this policy premarital screening for carriers and appropriate counselling are available.

***The Philippines*:** The only policies that can be related to genetics are:

- *UNICEF/UNDP/World Bank/WHO Special Programme for Research & Training in Tropical Diseases*[154]*;*
- *Operational Guidelines for Ethics Committees Reviewing Biomedical Research*[155]*;*
- *Ethical Guidelines for Genetic Research with a Section on Stem Cell Research.* (issued by the Philippine Council for Health Research and Development)[156];
- *The Intellectual Property Code of the Philippines* (Republic Act No. 8293)[157].

***South Africa*:** The *National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities* [158] document provides recommendations for the provision of genetic services. It has sections on general ethical guidelines for medical genetics and on ethical principles for genetic professionals.[159]

Ethical guidelines for genetic research purposes are available.[160] In the *National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities* the priority medical genetic services are described. These include services offered prior to conception, during pregnancy, at birth, in infancy and childhood, and in adolescence and adulthood. The way in which these services could be delivered at various levels from primary to tertiary health systems, is covered. The education of learners at schools and the training of genetic health professionals is addressed. Recommendations that medical geneticists' and genetic counsellors' posts should be provided, urgently, in every province in the country, in order to offer the services, have not yet been acted upon, and most provinces still have no posts at

[154] http://www.who.int/tdr/publications/documents/quality_practices.pdf (accessed May 2, 2013)
[155] Available at http://www.medicine.cmu.ac.th/research/ethics/OPGuide.pdf (accessed January 17, 2013)
[156] In: The Philippine National Ethical Guidelines for Health Research 2006. Available at https://webapps.sph.harvard.edu/live/gremap/files/ph_natl_ethical_gdlns.pdf. (accessed May 16, 2013)
[157] Available at http://www.chanrobles.com/legal7intellectualpropertycodeofthephilippines.html (accessed May 16, 2013)
[158] Department of Health, South Africa (2001). Policy Guidelines for the management and prevention of Genetic Disorders, Birth Defects and Disabilities. Department of Health, Pretoria, South Africa.
[159] Baumiller RC, Cunningham G, Fisher N, Fox L, Henderson R, Lebel R, McGrath G, Pelias MZ, Porter I, Seydel F, Wilson NR (1996). Code of Ethical Principles for Genetics Professionals. Am J Med Genet; 65: 177-183.
[160] Medical Research Council, South Africa (2002a). Guidelines on Ethics for Medical Research: General Principles. MRC South Africa, Tygerberg. Medical Research Council, South Africa (2002b). Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research. MRC, South Africa, Tygerberg.

all. Interventions are described, including strategies for prevention, such as genetic counselling, preconception and prenatal methods of prevention (e.g., PND), postnatal diagnosis and population screening. There are recommendations regarding: the integration of medical genetics laboratory services into the NHLS, which has been partially achieved, although co-ordination is still poor and duplications and inconsistencies still exist; the composition and functions of a Medical Genetics Advisory Board (it was suggested medical genetics professionals as well as a lawyer should be included); and the evaluation of human genetics programmes.

International conventions and directives, such as the *European Convention on Human Rights*, acknowledge that there are basic human rights for patients with genetic conditions and that everyone is entitled to basic health care.[161] In line with these international standards the South African Constitution of 1996 provides not only for fundamental rights such as the right to life (Section 11 of the constitution), to equality (Section 9), dignity (Section 10) and privacy (Section 14) but also provides, in Section 27, that everyone has the right to have access to health care, sufficient food, water and social security.

In terms of the *National Health Act* (NHA) of 2003[162] the State is obliged to provide free health care services to pregnant and lactating women and children under the age of 6 years. Furthermore, free PHC services must be provided to all those who are not members of medical aid schemes. Section 4 of the NHA also provides authority for women to have access to free termination of pregnancy, subject to the *Choice on Termination of Pregnancy Act 92* of 1996. This act provides the conditions and procedures to be followed for a person to obtain a termination of pregnancy. It states that a woman may obtain a termination upon request in the first 12 weeks, and thereafter, in consultation with a medical practitioner, where the health of the mother or the foetus may be at risk. The NHA (2003) has clear provisions in section 7 for consent to medical treatment and, with a few exceptions, a health care service may not be provided without informed consent. Further, section 8 of the NHA deals with the control of the use of blood products, tissue, and gametes and zygotes in humans and prevents the reproductive cloning of human beings.

[161] Convention on Human Rights and Biomedicine of the Council of Europe (1997).
[162] National Health Act 61 of 2003

Specific cultural and social issues pertaining to medical genetic services

Argentina**:** The main cultural and social issues pertaining to medical genetic services stem from the ***lack of genetic literacy among the population and the health professionals and the paucity of genetic services available throughout the country*****.**

Furthermore, ***coverage for genetic services by private and social insurance is very poor, which leads to discrimination and stigmatization of people with genetic disabilities*****.**

The lack of prenatal genetic services in the public sector and the prohibition of abortion for foetal reasons increase the stress and suffering of families affected with genetic conditions.

Low social class, low education level, and adherence to conservative religious precepts are associated with a high emotional burden from genetic defects.

Brazil**:** ***Poverty and lack of knowledge and understanding*** of genetic services, their purposes and what they can offer ***are the basic issues.***

Abortion is not viewed as an option by many, due to cultural and religious attitudes. However, some women do not consider the termination of a pregnancy of a malformed and non-viable foetus an abortion; thus abortion for specific medical indications would be acceptable to many.

Predictive and prenatal testing is culturally complicated, as the public is not used to taking the responsibility for decisions related to their own health and treatment. ***Many physicians who work in prenatal settings do not seem to understand the meaning of informed choice and autonomous decision making, and often make decisions for their patients.***
Participation of patients and informed autonomous decision making regarding health care is not a tradition in Brazil. Physicians are still viewed as holding all information regarding health, and for a long time, people have accepted being told what to do in this area. People making informed autonomous decisions about their health care has been changing in the last decade or so with the spread of access to communication and the internet supported by general education of the public. However, ***Brazil*** is still way behind in general medical education compared with other countries.

The importance of medical genetics and its integration into public health and prevention is not understood by most physicians, health authorities, public officials, and policy makers. Nor is the specialty of medical genetics known by many, who believe genetics will always be linked to very rare disorders, research,

laboratories, and high cost. Such beliefs are common amongst the lay public and health professionals. In consequence many patients/families are not referred to specialized services or only arrive in genetic services after recurrences, which could have been avoided.

China*:** There are a several specific cultural and social issues relating to genetic services in ***China. ***China*** consists of 56 ethnic minorities who differ in language, lifestyle, belief and culture. The distribution of congenital and genetic disorders may differ among specific groups and geographic locations.

(i) ***Language*** – the official language of ***China*** is mandarin, but only 53.1% Chinese can fluently communicate by mandarin. 86.4% are using the language with one of seven very distinct accents.[163] 53 ethnic groups have their own language. Most of the languages for small ethnic groups have distinct linguistic patterns, are rich in words for daily life, but lack scientific words.

(ii) ***Lifestyle*** – lifestyle of people also differs between ethnic groups.

(iii) ***Faith***– Buddhism, Christianity and Islam are present in ***China*** and Taoism represents also a large faith group. Some ethnic groups may also have their own faith.

(iv) ***Traditional Chinese medicine*** – ***Chinese herbal medicine is deeply rooted in the Chinese culture***. Thus most Chinese still consult both Western medical health professionals and traditional Chinese herbal medical practitioners.

All these issues can affect the ways in which genetic services are delivered and received, the communication and interactions in genetic counselling sessions and the choices people make.

Egypt*:** In general, there is still ***lack of awareness concerning the importance of the services for early intervention and prevention of genetic and congenital disorders.

Misconception about some services include the mistaken idea by some that antenatal genetic testing is merely a step used by women to obtain abortion for a unwanted pregnancies.

Fixed cultural and social beliefs and fear of stigmatization are common issues that prevent people from asking for such genetic services.

[163] Data provided by Nanbert Zhong, Peking University Center of Medical Genetics, Beijing, People's Republic of China.

This is especially common among the rural population, mainly the rural communities of the southern part of Upper Egypt. This might be attributed to the ***high prevalence of illiteracy and customary consanguineous marriages*** amongst these people.
There is a great discrepancy between rural and urban population as regards their perception of intervention and preventive approaches to congenital and genetic disorders. Changing such attitudes in rural populations (57% of the total population) is difficult, but modifying their to date fixed cultural and social beliefs will be essential to enable rural populations to gain maximum advantages from utilizing available community genetic services.

India is a vast country with great social, cultural, religious and ethic diversity.
Consanguineous marriages are practiced by most communities in south ***India***, the frequency ranging from 20 – 30 % of unions. This occurs both among the Muslim and Hindu communities. In north ***India*** consanguineous marriages are practised mostly by Muslims.[164] However in the rural areas a small number of consanguineous marriages, 3-5 %, are seen in the Hindu community.[165]
In addition, throughout ***India, many people marry within their own ethnic group (endogamy)***, although not consanguineously. This results in the presence of founder mutations.
These practices result in a higher birth prevalence of autosomal recessive disorders and numerous founder mutations in the Indian population. Many disorders are common among certain communities (**Box 3.9**).

Stigmatization of those carrying mutant genes (especially women). The stigma associated with being a thalassaemia minor carrier has been well documented by social studies. For instance, a study conducted in Bengal observed *"that blood is deeply valued in the Bengali Kinship system and this genetic mutation is perceived to be corrupting the blood. Being a thalassaemia carrier (i.e. having thalassaemia minor) renders an individual unfit as a suitable marriage partner because of beliefs related to purity of blood, its association with the continuity of the lineage, and subsequent transmission of desirable traits to future generations. The risk of non-marriage affects women disproportionately, and parents are not inclined to test their daughters because of the possibility of not being able to marry them off to eligible suitors."*[166]
Such data highlight the need for genetic counselling to take into account the perception of the tribal communities and to develop anthropological tribal oriented approaches to avoid stigmatization.

The possibility of stigmatization of women found to be carriers of X-linked disease also needs careful consideration in genetic counselling consultations.

[164] Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.
[165] Data provided by Ishwar C. Verma, Centre of Medical Genetics, Sir Ganga Ram Hospital, Rajender Nagar, New Delhi, India.
[166] Verma IC, Saxena R, Kohli S: Past, present & future scenario of thalassaemic care & control in India. Indian J Med Res 134, October 2011, pp. 507-521.

Oman*:** The ***custom of consanguineous and arranged early marriages in Muslim communities is deeply rooted in Arab culture. The balance of opinion in the Middle East still remain in favour of consanguinity irrespective of increased risk of autosomal recessive diseases, congenital malformations and mental retardation.
Recent data suggest that there is a reduction in the frequency of consanguineous marriages particularly in urban areas of ***Oman***.
The present policy to address the issue includes premarital identification of carrier risk by genetic screening and counselling of carriers to afford them the opportunity of deciding not to marry.

***The Philippines*:** Several cultural and social issues affect the delivery of genetic services.

(i) ***Termination of pregnancy is illegal:*** termination of pregnancy is not available and is considered illegal by law; this is due to a largely Catholic influence in the country.

(ii) ***Religious beliefs and traditional practices*** affect the way people view health and the causes of disease. Those living in far-flung provinces would still seek help from traditional faith healers rather than consult a medical professional.

(iii) ***Lack of education and understanding of basic genetic concepts:*** Although genetic awareness is rising among the Filipinos, the lack of education and understanding of basic genetic concepts, also contribute to the misconceptions about genetics.

(iv) ***Infections remain to be the top priority of the DoH.*** The majority of programmes and strategies are designed to combat communicable diseases. With the increasing awareness among Filipinos of the different genetic disorders, the demand for genetic services however has increased throughout ***the Philippines.***

(v) ***Provision of basic genetic healthcare services to every region remains the biggest challenge.*** While it would be preferable to have at least one geneticist and one genetic counsellor in each region, this is currently not possible. ***There are only a few geneticists with clinical practices, available*** only in the major urban areas such as Manila, Cebu, and Davao. In response to this ***critical lack of specialists***, the DoH and the NSRC offer scholarships for fellowships in clinical genetics for paediatricians committed to practicing clinical genetics in regions currently without services. In addition, a MSc programme in genetic counselling was established in 2011 (see “Genetic counsellors as a recognised and registered profession”).

South Africa*:** There are a several specific cultural and social issues relating to genetic services in ***South Africa[167]***:***

(i) ***Systems of thought***, prevailing fatalistic attitudes, communal decision-making, the indistinct line between life and death, and belief in the power of ancestral spirits.

(ii) ***Beliefs and myths*** about the causes of genetic disorders.

(iii) The tendency, in the majority of people, to ***consult both western medical health professionals and traditional healers***.

(iv) ***The custom of consanguineous marriage***, common practices and taboos.

(v) ***Language and communication***, since in most local languages there are no terms for words such as genes and chromosomes.

All these issues can affect the ways in which genetic services are delivered and received, the communication and interactions in genetic counselling sessions and the choices people make. They are integral to an issue prevalent in other countries, namely a lack of genetic literacy among the population and the health professionals and the paucity of genetic services available throughout the country.

[167] Kromberg JGR, Jenkins T (1997). Cultural Influences on the Perception of Genetic Disorders in the Black Population of Southern Africa. In Clarke A, Parsons E (eds). Culture, Kinship and Genes. London, Macmillan, 147-157.

Assessment of the attention given to medical genetic services by the national government/policy makers as compared to other health issues

In ***Argentina*****,** genetic services **do not receive sufficient** attention by national government/policy makers as compared with other health issues.

Brazil**: Genetic services are not well recognized both by the general public and the medical profession.** Education and literacy for the public and medical profession in genetics is needed.

China**:** The central government has given a great deal of attention to maternal and child health since the founding of the People's Republic in 1949. In the last two decades, ***China*** has made substantial progress in reducing maternal, infant and under-five mortality. Most of the policies related to medical genetics were developed for birth defect control by the maternal & child health division of the Central MoH. For example, the policy of providing free folic acid for the prevention of neural tube defect renewed in 2010, the regulation of technology for foetal chromosome karyotyping issued in 2008. However, there is no regulation regarding molecular genetic diagnosis.

Egypt**:** To date **not much attention has been given to the genetic services** from health policy makers. Providing genetic services is not a priority in the new health reform system (HRS). The key priority in the HRS is to achieve universal insurance coverage for all Egyptians.
The MoH&P has focused on other health issues including the avian flu and H1N1 flu (Swine Flu). Most of the attention was directed to these two top priority health crises. Budgets were reallocated and even health care professionals were relocated to participate in surveillance and prevention of the epidemic infection.
Currently the family planning programme, the expanded childhood immunization programme, HIV, Hepatitis C and cancer prevention and treatment are the main interest of the MoH&P and are priority health issues.

India**:** The national government has shown great interest in non-communicable disorders, such as cardiovascular diseases, diabetes mellitus, stroke, chronic lung disorders and cancer. The pilot phase of the *National Program for Prevention and Control of Cancer, Diabetes, CVD and Stroke* (NPCDCS) for these disorders was launched in January 2008, with an outlay of Rs. 1660 crores (~233 million €/ 305 million US$ in April 2013) in the 11th 5-year plan.[168] **Unfortunately the NPCDCS programme does not include congenital and genetic disorders.** Although talks for initiating a nationwide programme for the care and prevention of

[168] http://health.bih.nic.in/Docs/Guidelines-NPCDCS.pdf (accessed April 30, 2013)

haemoglobinopathies have been ongoing for several decades, no concrete national programme has yet been started by the government. The ICMR has now taken the lead to start a *"National Haemoglobinopathies Control Programme"*.[169] It will start in Delhi, Chandigarh and Punjab as a pilot study (**see Box 3.6**). The ICMR has also taken the initiative to establish five additional regional haemoglobinopathy centres at medical colleges in Maharashtra, Gujarat, West Bengal, Karnatakar and Punjab for molecular and prenatal diagnosis.
In February 2013, a new health initiative *"Rashtriya Bal Swasthya Karyakram"* was launched by the government. The initiative is set to provide comprehensive health care *"and improve the quality of life of children through early detection of congenital disorders, diseases, deficiencies, development delays including disability"*.[170]

***Oman*:** Increasing attention has been given to medical genetic services in the past years. Since the implementation of the *7th Five-Year Plan for Health Development* (2006-2010) by the MoH, the Sultanate of Oman has a national strategic plan on congenital and genetic diseases. (see "Existing national policies, guidelines and planning activities for the provision of medical genetic services")

***The Philippines*:** Aside from newborn screening, the DoH has supported (financially) the *Philippine Birth Defects Surveillance (PBDS) Project*[171], the *TRS Project* and the *Preconception Health (PH) Project*, in collaboration with the IHG-NIH-UP. The PBDS is currently implemented in 18 sentinel sites (different political/geographical regions) with 82 health facilities and communities. The TRS is currently implemented in 10 sites, and the goal is to make genetics services accessible to all patients with congenital and genetic disorders through the use of a web-based referral system. Aside from these projects, there is very little attention provided for the other aspects of medical genetic services.
The future of genetic services is dependent on a variety of factors. Limited attention is provided by government since the focus is still on eradication of infectious diseases that predominate the top ten causes of infant mortality and infant morbidity.

Learning from the developed countries where eradication of infections eventually paved the way to improvement of genetic services, ***the Philippines*** must prepare now by giving more attention (in terms of budget and programme planning) to congenital and genetic disorders.

[169] Colah, R: Control, Strategies for Hemoglobinopathies in India. 1st Pan-Asian Conference on Haemoglobinopathies, country reports. IThalassemia Report 2012; 2 (s1), p.1-2.
[170] Press Information Bureau, Government of India, 27 March, 2013. Online available at http://pib.nic.in/newsite/erelease.aspx?relid=92045 (accessed April 12, 2013)
[171] Padilla CD et al.: Establishment of the Philippines Birth Defects Surveillance. Acta Medica Philippina. 2011. Vol. 45, No. 4. Available at http://actamedicaphilippina.com.ph/sites/default/files/vol_45_no_4_fulltxt.pdf (accessed May 13, 2013)

***South Africa*:** The national policy regarding medical genetic services, which was set out in the *National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities*, was developed before the impact of the HIV/ AIDS pandemic became apparent. That epidemic together with other problems, including the increasing incidence of tuberculosis (TB), poor governance and difficulties in health service delivery, has resulted in medical genetic services having diminished priority compared with the 1990s. It is hoped that this situation will improve consequent on the recognition, by the WHO, that congenital and genetic disorders present major health problems, and, in 2010, their recommendation that services for the care and prevention of congenital and genetic disorders in developing countries should be prioritised.[172]

[172] World Health Organization (2010). Sixty-Third World Health Assembly. Provisional agenda item 11.7. April 2010. Available at: http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf (accessed May 16, 2013)

In conclusion

With the probable exception of ***Oman*** the national governments of the other GenTEE countries have only given **limited attention to legislation and regulation of services** for the care and prevention of congenital and genetic disorders and still **need to develop strategies to strengthen their genetic services** and to enable their primary care services to use the services for the benefit of their patients. It is obvious that in large countries like ***India***, the introduction of services for population screening programmes (e.g. for haemoglobinopathies) will be a gradual process.

Despite most of GenTEE countries progressing well through epidemiological transition the realisation that this increases the public health significance of congenital and genetic disorders has not been fully recognized. **Genetic testing services for chronic diseases with subgroups with significant genetic risk components non-communicable chronic diseases such as: heart disease, stroke, cancer and diabetes are hardly available.** The WHO has indicated over the last decade the need for middle- and low-income countries to consider the need for medical genetic services. Congenital and genetic disorders and services for their care and prevention were confirmed as a global priority, particularly in middle- and low-income nations by the WHO's World Health Assembly in 2010.

South Africa is a special case. The Constitution and laws protect the rights of the disabled and at the turn of the 21st century it had put in place very progressive thinking national guidelines for genetic services. However, for several reasons, including a change of priorities because of the HIV/AIDS and the TB epidemics, the policies still wait for full implementation.

VII Research priorities in genetics/genomics

Argentina

In ***Argentina***, biomedical research has a long tradition in academic centers.

The *Consejo Nacional de Investigaciones Científicas y Técnicas* (CONICET, National Council for Scientific and Technological Research[173]) has existed for over 50 years, albeit with changing roles. Its main function today is to fund and administer a researcher career. CONICET, in turn, is part of the Argentinian Ministry of Science, Technology and Productive Innovation, which in 2009 started a concerted effort to develop public-private partnerships for research and development in genomic approaches in biotechnology and biomedicine. Most scientific and technical research is conducted at Argentinian (public) universities with public and private funds.

The National MoH has a lesser role in genetic/genomic research.[174]

Genetics/genomics research is conducted in several centres, such as the:

- Department of Molecular Biology of the University of Buenos Aires[175];
- Instituto de Investigaciones de Ingenieria Genetica y Biologia Molecular[176];
- Fundacion Leloir[177];
- Instituto Multidisciplinario de Biología Celular[178], and the
- Instituto de Biología Molecular y Celular of the Universidad Nacional de Rosario[179].

Specific areas of genetics/genomics research are:

- basic molecular genetics;
- immunogenetics;
- molecular population genetics;
- forensic genetics;
- genome sequencing (the *Trypanosoma cruzi* genome was sequenced by a multinational team in which Argentine geneticists from two separate institutions played a key role);
- gene therapy;
- cancer genetics and
- stem cell research.

[173] www.conicet.gov.ar (accessed May 16, 2013)
[174] www.saludinvestiga.org.ar (accessed May 16, 2013)
[175] http://exactas.uba.ar (accessed May 16, 2013)
[176] www.ingebi-conicet.gov.ar (accessed May 16, 2013)
[177] www.leloir.org.ar (accessed May 16, 2013)
[178] www.imbice.org.ar (accessed May 16, 2013)
[179] www.ibr.gov.ar (accessed May 16, 2013)

Research in clinical genetics is concentrated in dysmorphology–cytogenetics, selected single-gene disorders such as skeletal dysplasias, muscular dystrophies, fragile X syndrome, CF, thalassaemia, congenital deafness, and cancer.[180]

In 2009 the National MoH convened a large group of geneticists conducting research to discuss priorities in genetics research. However, the results of this exercise have not been disseminated nor acted upon.

Research funding by private parties in Argentina

Research funding for genetics by private parties is very scarce in ***Argentina***.

Known co-operations with international funding agencies in Argentina

There are a number of research projects in basic genetics that have received funding from several international bodies, such as the USA National Institutes of Health (NIH), Fogarty Center, CDC, Howard Hughes Foundation, Wellcome Trust and others. None of these agencies fund clinical projects.

Brazil

Brazil has made significant investments to fund research in medical genetics and genomics. The main funder is the public sector, both state and federal.
At the federal level, the major funders are the:

- *Financiadora de Estudos e Projetos (FINEP)* [Financier of Studies and Projects[181]],
- *Coordenação de Aperfeiçoamento de Pessoal de Nível Superior (CAPES)* [Coordination of Improvement of Higher Education Personnel[182]] and the
- *Conselho Nacional de Desenvolvimento Científico e Tecnológico (CNPq)* [National Council for Scientific and Technological Development[183]].

FINEP is acting under the Ministry of Science and Technology (MCT)[184], which focuses on fostering the institutional projects (universities, companies, and institutes of technology).

CAPES invests in training high-level human resources in the country and abroad.
CNPq is an agency of the MCT for the promotion of scientific and technological research and training of human resources for research in the country. It focuses on

[180] Penchaszadeh VB (2008). Argentina: Public Health Genomics. Public Health Genomics;12:59-65.
[181] www.finep.gov.br (accessed May 16, 2013)
[182] www.capes.gov.br (accessed May 16, 2013)
[183] www.cnpq.br (accessed May 16, 2013)
[184] www.mct.gov.br (accessed May 16, 2013)

encouraging researchers and their projects, individually or in groups. CNPq, in particular, has supported specific actions in clinical genetics through partnerships with the National MoH.[185]

Among the projects, several were structured partnerships with institutions in several states of ***Brazil***, forming various networks such as *Familial Cancer Network*, the *Network for Diagnosis in Inborn Errors of Metabolism (Rede-EIM-Brasil)*, and others.

At the state level major funders are the:

Fundação de Amparo a Pesquisa (FAP) [Foundations for Research Support] in each state of ***Brazil***. One of these FAPs, the *Fundação de Amparo à Pesquisa do Estado de São Paulo, FAPESP* (Foundation for Research Support of São Paulo)[186], is the fourth largest funder of scientific and technological development of the country. In 2009 FAPESP grant aid totalled nearly $ 500 million. A major achievement of FAPESP was the establishment of genomic research in the country, starting with an agreement with the ONSA network (*Organization for Nucleotide Sequencing and Analysis*) in 1997.[187] These efforts resulted in the sequencing of the genome of *Xylella fastidiosa* in 2000[188] and established an expert network on advanced projects in genomics, with international impact. Its most important project in human genetics was the *Human Cancer Genome Project* (2011). In this project, about 2 million DNA sequences of normal and tumour tissue were deposited in GenBank[189]. Other projects, for example the *Clinical Genome Cancer Project*[190], followed.

Another form of support is the partnership of different funders (CNPq, CAPES, FAP, National MoH, Ministry of Education, and others) by joint financing projects, like the programme of the *Institutos Nacionais de Ciencia e Tecnologia* (INCT) (National Institutes of Science and Technology 2011). Released in July 2008, this programme has established itself as a powerful instrument for advancing science, technology, and innovation in the country. With over a hundred projects approved in different research areas, such as health, biotechnology, nanotechnology, and energy, the programme aims to mobilize and aggregate in networks the best research groups in frontier areas of science and in strategic areas for the sustainable development of the country.

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[185] The partnership is based on edicts: the MCT / CNPq / MS-SCTIE-DECIT 21/2006 edict (http://portal.saude.gov.br/portal/files/pdf/genetica.pdf, accessed January 29, 2013) and MCT / CNPq / CT-HEALTH No. 57/2010 (www.cnpq.br/editais/ct/2010/057.htm, accessed January 29, 2013) which aimed to support structuring projects for health care in Medical Genetics throughout Brazil

[186] www.fapesp.br (accessed May 16, 2013)

[187] Simpson AJ, Perez JF. (1998). ONSA, the São Paulo Virtual Genomics Institute. Organization for Nucleotide Sequencing and Analysis. Nat Biotechnol 16:795-6.

[188] Simpson, AJG, Reinach FC, Arruda P, Abreu FA et AL. (2000). The genome sequence of the plant pathogen Xylella fastidiosa. Nature 406: 151-157.

[189] Brentani H, Caballero OL, Camargo AA, da Silva AM et al. (2003). The generation and utilization of a cancer-oriented representation of the human transcriptome by using expressed sequence tags. Proc. Natl. Acad. Sci USA 100: 13418-13423.

[190] Wünsch-Filho V, Eluf-Neto J, Lotufo PA, da Silva Jr. WA and MA Zago. (2006). Epidemiological studies in the information and genomics era: experience of the Clinical Genome of Cancer Project in São Paulo, Brazil. Braz J Med Biol Res 39: 545-553.

The following institutes are linked to the further development in human genetics and medicine:

- National Institute of Science and Technology Cell Therapy, based in the Faculty of Medicine of Ribeirão Preto University of São Paulo[191];
- National Institute of Science and Technology of Stem Cells in Human Genetic Diseases, based at the Institute of Biosciences, University of São Paulo[192];
- National Institute of Science and Technology for Cancer Control, based at the National Cancer Institute[193] (linked to the National MoH[194]);
- National Institute of Population Medical Genetics[195], based at the Hospital de Clinicas de Porto Alegre–Federal University of Rio Grande do Sul [196] (INAGEMP 2011);
- National Institute of Science and Technology of Molecular Medicine, based at the Faculty of Medicine, Federal University of Minas Gerais[197]; and the
- National Institute of Science and Technology in Oncogenomics, based at the Cancer Hospital of São Paulo[198].

Research funding by private parties in Brazil

Some pharmaceutical and biotech companies are funding clinical research in the field of new drugs, mostly phase 3 and 4 studies, especially for rare diseases.

Known co-operations with international funding agencies in Brazil

The *Human Cancer Genome Project* [199]. In this project, about 2 million DNA sequences of normal and tumour tissue were deposited in *GenBank*[200] and used for other projects as the *Clinical Genome of Cancer Project*.[201]

[191] www.fmrp.usp.br (accessed May 16, 2013)
[192] www.ib.usp.br (accessed May 16, 2013)
[193] www.inca.gov.br (accessed May 16, 2013)
[194] www.saude.gov.br (accessed May 16, 2013)
[195] www.inagemp.bio.br (accessed May 16, 2013)
[196] www.hcpa.ufrgs.br (accessed May 16, 2013)
[197] www.medicina.ufmg.br (accessed May 16, 2013)
[198] www.accamargo.org.br (accessed May 16, 2013)
[199] www.compbio.ludwig.org.br/ORESTES (accessed May 16, 2013)
[200] Brentani H, Caballero OL, Camargo AA, da Silva AM et al. (2003). The generation and utilization of a cancer-oriented representation of the human transcriptome by using expressed sequence tags. Proc. Natl. Acad. Sci USA 100: 13418-13423.
[201] Wünsch-Filho V, Eluf-Neto J, Lotufo PA, da Silva Jr. WA and MA Zago. (2006). Epidemiological studies in the information and genomics era: experience of the Clinical Genome of Cancer Project in São Paulo, Brazil. Braz J Med Biol Res 39: 545-553.

China

In ***China***, government funding is available, mainly through the *Natural Science Foundation of China* (NSFC)[202] and the Ministry of Science and Technology. The full funding scheme consists of three groups:

(i) research projects (funding is limited to individual research topic);
(ii) research scientist developmental project (funding is limited to a person or a group for a specific research);
(iii) environment condition projects (funding is limited for improving laboratory's hardware condition, e.g. equipment).

Medical genetic research is eligible for funding from all three funding schemes.
Apart from the funding schemes provided nationally by the central government, each province, autonomous region and municipality also offers funding for encouraging research locally.

During the last decade, the central government has undertaken concerted efforts to move the country into the upper echelon of genetic and genomic research world-wide. In 1998, the Ministry of Science and Technology[203] established the *Chinese National Human Genome Centre* (CHGC) in Beijing[204] and Shanghai[205], and in 1999 the *Beijing Institute of Genomics* (BGI)[206] as centres of excellence for genome sequencing and analysis. The establishment of these institutes enabled ***China*** to participate in the *Human Genome Project* and to contribute to the International *Human HapMap Project*.

China is also a partner in the *International Cancer Genome Consortium* (ICGC).

Since the establishment of two large-scale population-based biobanks: the *Kadoorie Study of Chronic Disease in China* (KSCDC) and the *Guangzhou Biobank Cohort Study* (GBCS), ***China*** is in the process of setting up large-scale population-based biobanks. ***China's*** diverse population of 56 different ethnic minorities are of special interest in regard to genetic disorders but also in regard to hereditary evolution. Understanding of the genetic bases of chronic diseases reflects the changing morbidity and mortality pattern caused by ***China's*** epidemiological transition. A new initiative of longitudinal cohort study for pregnancy outcome has promoted a biobanking of pregnancy-related specimens that may allow researchers to investigate the nature of pregnancy and its outcomes on a long term basis.

[202] www.nsfc.gov.cn (accessed May 16, 2013)
[203] www.most.gov.cn (accessed May 16, 2013)
[204] www.chgb.org.cn (accessed May 16, 2013)
[205] http://chgc.sh.cn/en (accessed May 16, 2013)
[206] www.genomics.cn/en (accessed May 16, 2013)

Research funding by private parties in China

Research funding received from various private sources is available. Most of funding is bound to an operative body, generally a university, for a short term. The universities review and fund acceptable research projects for their own staff and students. Unlike in the Western countries, pharmaceutical companies in ***China*** have almost none investment supporting research. Family or individual-driven associations for a particular disease, are a limited funding resource due to limited fund raising opportunities for this type of initiative.

Known co-operations with international funding agencies in China

Chinese researchers in human genetics collaborate with international research teams and have received international funding from bodies such as the NIH, Fogarty Foundation, WHO, Bill & Melinda Gates Foundation, Welcome Trust, CDC, MoD and the EU FPs.

Egypt

In *Egypt*, research in human genetics/genomics is primarily done at the NRC, the largest research and development centre in ***Egypt***[207]). In addition, research in genetics along with other research and surveys under the theme "prevention of disabilities" is included within the yearly plan for research presented by the MoH&P's Children with Special Needs Department.

Research funding by private parties in Egypt

International pharmaceutical companies fund pharmacogenetic related research. Other private companies (e.g. Clinilab and Genzyme) provide equipment and drugs for research purpose.

Known co-operations with international funding agencies in Egypt (some data are unavailable):

(i) The EU funded project with the Kasr El-Einy friends association is researching prevention, early detection and early intervention in Egyptian children with genetic disability and children at risk. Ten metabolic disorders are screened using tandem mass (MS/MS technique) for screening and diagnosis. Filter paper stored blood specimens from 25000 newborns is utilized in screening for CH.

[207] www.nrc.sci.eg (accessed May 16, 2013)

(ii) A EU funded project is screening for PKU and Galactosemia on 60.000 filter paper neonatal blood specimens previously used in neonatal screening for CH. The specimens were acquired in the three main zones in Alexandria.

(iii) EUMEDIS (*Euro-Mediterranean Information Society*)[208] project; MEDGENET (Euro-Mediterranean Network for Genetic Services[209]). A EU funded project in collaboration with the European Genetic Foundation, Bologna, Italy.

India

During the last decade, substantial funds have been made available by the DST[210], DBT[211], CSIR[212] and the ICMR[213] to fund genetics/genomics research. All these organizations have identified priority areas for funding especially in the area of infectious diseases, but including non-communicable disorders.

In 1998, the *Functional Genomics Unit* was established at the *Center for Biomedical Technology*, later renamed *CSIR Institute of Genomics and Integrative Biology* (IGIB), focussing mainly on genomics and bioinformatics[214]. In 2009, the IGIB mapped the human genome for the first time in ***India***.

In 2009, the government approved the establishment of the *National Institute of Biomedical Genomics*[215] (NIBMG) as an autonomous institution. This is the first institution in ***India*** devoted specifically to capacity building in biomedical genomics, and to conduct of basic, clinical and translational research in biomedical genomics in an interdisciplinary and integrated way. NIBMG will house an interdisciplinary infrastructure that is required for frontier research and applications in biomedical genomics, and for training students and scientists in medical-genetic research, translation and service.

The Indian government has declared 2010-2020 as the *"Decade of Innovation".* The programme will be focusing on developing new capabilities in emerging areas such as genomics and biotechnology arguing "*The sequencing of the human genome in India by Institute of Genomics and Integrative Biology (IGIB), Delhi, a constituent laboratory of Council of Scientific and Industrial Research (CSIR) has helped India join the league of select countries undertaking advanced research in the area of genomics."*.[216]

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[208] http://www.eumedis-cy.ucy.ac.cy/initiative.html (accessed May 16, 2013)
[209] http://cordis.europa.eu/search/index.cfm?fuseaction=proj.document&PJ_RCN=8823573 (accessed May 16, 2013)
[210] www.dst.gov.in (accessed May 16, 2013)
[211] http://dbtindia.nic.in/index.asp (accessed May 16, 2013)
[212] www.csir.res.in (accessed May 16, 2013)
[213] www.icmr.nic.in (accessed May 16, 2013)
[214] http://www.igib.res.in/ (accessed May 16, 2013)
[215] www.nibmg.ac.in (accessed May 16, 2013)
[216] Sanaa Arora: India 2010:2020 – Decade of Innovation. Online available at www.strategicforesight.com/india_2010_2020.htm (accessed May 16, 2013)

Like ***China***, ***India*** is part of the ICGC.[217] ***India*** is focussing on oral cancer, as this cancer is common in the Indian population.

Another international project is the *Stanford India Bio-Design* that aims at training the next generation of medical technology innovators in ***India***.[218]

India is determined to prevent foreign bio piracy of human bio resources. National guidelines not only require Indian DNA samples to be analysed by national scientists in national laboratories but also forbids the transfer of DNA samples out of the country.[219]

Research funding by private parties in India

Some of the pharmaceutical companies and biotech companies are funding research in the field of genomics for drug discovery. The Chatterjee Group, an investment company in the USA, through its Institute of Molecular Medicine has set up a Centre of Genomic Application in Delhi and a Centre for Population Genomics in Kolkata.

Known co-operations with international funding agencies in India

A large number of projects in various medical institutions and universities are being carried out in collaboration with NIH, CDC and other universities in USA, UK, and Europe. There are a number of projects funded by the EU.

Oman

In ***Oman***, funds are offered from the TRC[220] which prioritises projects benefiting the local community. Furthermore, SQU[221] funds small and medium –size research programmes, and His Majesty Sultan Qaboos' Research Fund offers substantial research grants.

Research funding by private parties in Oman

Private companies offer limited research grants

Known co-operations with international funding agencies in Oman

The Human Genetic Unit of the MoH[222] collaborates with centres of excellence in Germany, the Netherlands, UK and USA as well as the SQU.

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[217] www.icgc.org (accessed May 16, 2013)
[218] http://biodesign.stanford.edu/bdn/india/ (accessed May 16, 2013)
[219] Sleeboom-Faulkner M (ed.) (2009) Human Genetic Biobanks in Asia. Oxford: Routledge.
[220] www.trc.gov.om (accessed May 16, 2013)
[221] www.squ.edu.om (accessed May 16, 2013)
[222] www.moh.gov.om (accessed May 16, 2013)

The Philippines

In ***the Philippines***, genetics and genomics compete with other disciplines in securing funding for research grants. Genetic and genomic research continue to compete with the top 10 causes of morbidity and mortality for available research funds. The DoH is offering funding opportunities for operational research in genetics. The Department of Science and Technology has dedicated funding support for the Philippine Genome Center (PGC) but limited its use to certain diseases (i.e. neglected tropical diseases, cardiovascular diseases and diabetes).
A new development (since January 2013) is that the government will, through a special government budget of the CHED and with the creation of the Philippine California Advanced Research Institutes (PCARI), support innovative health and translational research.
The IHG-NIH[223] is the major public research institute for both congenital and genetic disorders and complex diseases.

Research funding by private parties in the Philippines

There is very limited funding from private parties. Some pharmaceutical companies involve local researchers in international projects.

Known co-operations with international funding agencies in the Philippines

There are several collaborations being undertaken by researchers, especially for complex diseases. Two current collaborations are:

(i) the PBDS Project[224] – supported by MoD *Global Network for Maternal and Infant Health;*

(ii) sub-phenotyping and genetics in oral-facial cleft families in the Philippines – in collaboration with the University of Iowa (USA) and the University of Pittsburgh (USA).

South Africa

In ***South Africa***, there are no policies specifically covering funding for research in human or medical genetics/genomics. However, the government does fund some medical research, including research in the field of human genetics/genomics, through two bodies, the *SA Medical Research Council (SAMRC)*[225] and the *National Research Foundation (NRF)*[226].

[223] http://ihg.upm.edu.ph (accessed May 16, 2013)
[224] http://ihg.upm.edu.ph/index.php?option=com_content&view=article&id=57&Itemid=58 (accessed May 16, 2013)
[225] www.mrc.ac.za (accessed May 16, 2013)
[226] www.nrf.ac.za (accessed May 16, 2013)

Through the NRF the National Department of Science and Technology (NDST)[227] has a programme that financially augments certain research grants and this has aided medical genetics/genomics research. The NDST, through its biotechnology strategy has funded two *Biotechnology Regional Innovation Centres (BRICS)*. These are high throughput genomics laboratories, namely the *LifeLab* at the University of KwaZulu Natal[228] and *Centre for Proteomics and Genomic Research* at the University of Cape Town[229], in the Western Cape Province. In the former laboratory work is mainly on infectious diseases and in the latter contract research in the field of human genetics/genomics is undertaken. The NDST also funded the *National Bioinformatics Network* and, recently, has provided funding for 2010 and 2011 for Phase 1, the planning phase, of a *National Human Genome Initiative*. The NHLS[230] Research Trust[231] funds research in pathology, including medical genetics. Staff across the NHLS received 367 research grants during the 2008-2009 year, valued at ZAR 124 million (~10.6 million €/ 13.9 million US$ in April 2013), with ZAR 21 million (~1.8 million €/ 2.3 million US$ in April 2013) coming from the NHLS Research Trust.[232] The amount awarded specifically for human genetics research is not available. However, projects in human genetics receive funding (on a competitive basis), every year, as well as some long-term funding.

Research funding by private parties in South Africa

Research funding is received from various private sources for short term projects (generally) on an ad hoc or regular basis. The universities screen and fund research projects for their own staff and students. Further, several private donor research foundations (e.g. Richard Ward Foundation at WITS University) held by universities have funded genetics projects from time to time. Some of the funding for human genetics projects at the University of Cape Town comes from genetic support groups, such as *Retina South Africa* who have funded research on inherited retinal disease, over many years, and the *Muscular Dystrophy Foundation* who fund various research projects in their field. Also, the *Cancer Association of South Africa (CANSA)* funds research on cancer at several universities.

Known co-operations with international funding agencies in South Africa

South African researchers in human genetics collaborate with international research teams and have received international funding from bodies such as the NIH, Fogarty Foundation, WHO, Wellcome Trust, US Aid, CDC, the MoD and the EU through the EU FPs. Also, the *Genographic Project* of the National Geographic Society has supported (2006-present) population genetic studies in the Human Genome Diversity and Disease Research Unit, at WITS University.

[227] www.dst.gov.za (accessed May 16, 2013)
[228] www.ukzn.ac.za (accessed May 16, 2013)
[229] www.cpgr.org.za (accessed May 16, 2013)
[230] www.nhls.ac.za (accessed May 16, 2013)
[231] www.nhls.ac.za/?page=nhls_research_trust&id=32 (accessed May 16, 2013)
[232] National Health Laboratory Service (2009) Annual Report 2008–2009. NHLS, Johannesburg

Current centres of excellence in genetics/genomics research

Currently centres of excellence in genetics/genomics research are present in most of the countries, apart from ***South Africa***. However, approximately 10 years ago the SAMRC[233] awarded 3 MRC of South Africa Human Genetics Research Units. These units of excellence are at WITS University[234] (Human Genome Diversity and Disease Unit), the University of the Western Cape[235] (Unit for Capacity Development in Bioinformatics), and the University of Cape Town[236] (Human Genetics Research Unit). These units are focused on capacity development.

In conclusion

In contrast to largely underdeveloped, underfunded genetic services in the public domain, the governments of most GenTEE countries have put substantial resources into genetic, mainly genomic, research during the last decade. This includes ***Brazil***, ***China***, ***India***, ***South Africa***, the purpose being to promote research **with the aim to become self-reliant in frontline research areas**.

Even lower-middle-income countries have committed funds into furthering genomic research as in ***the Philippines*** where the government founded a national genome centre.

Countries such as ***Brazil***, ***China*** and ***India*** clearly pursue the **strategic goal to move their countries into the vanguard of genomics research**. This often addresses research outside the field of medical genetics, and includes oncogenomics, pharmaceutics, communicable disease control, vaccine development and pharmacogenomics.

Although ***Brazil***, ***China*** and ***India*** have developed cutting-edge capacity in a remarkably short period of time to undertake genetic/genomic research, **huge gaps exist in the translation of such research into routine health services** due to the **lack of capacities in the health care sector** and poorly developed genetic services policy and infrastructure in the public domain. As a result, **in most GenTEE countries genomics research is not connected with public health services.**

[233] www.mrc.ac.za (accessed May 16, 2013)
[234] www.wits.ac.za (accessed May 16, 2013)
[235] www.uwc.ac.za (accessed May 16, 2013)
[236] www.uct.ac.za (accessed May 16, 2013)

VIII Patient organizations and public education in genetics

Establishment of patient organizations, main activities including lobbying and advocacy activities

Argentina has a long tradition of patient organizations as advocates for services and research and the number and strength of patient organization is growing.

Most patient organizations link with health professionals and research scientists to stimulate the development of new treatments and prevention strategies.
While most patients' organizations deal with a particular condition, there has been recently a tendency toward union in umbrella groups. One such umbrella group is the *Geiser Foundation*. [237]

Due to lobbying activities of patient organizations, a number of laws have been passed in Congress for the support of patients with specific conditions and research.
The National Ministry of Science and Technology has funded a research project on the needs of patients and families with rare diseases, which detailed those needs and presented them to Congress in 2009.[238]

Brazil: There are at least 100 different organizations, some just plain "kitchen table" and very few more professionally organized, such as the *Associação Brasileira de Assistência a Mucoviscidose* (ABRAM).[239]
It is very hard to access data on how many organizations for genetic disorders exist in ***Brazil***. Data provided by the *Aliança Brasileira de Genetica* (ABG[240]) show there are 40 affiliated organizations. But this is certainly underestimated, as the *Federação Brasileira das Associações Síndrome de Down*[241] is not part of the ABG, and congregates more than 60 associations of Down syndrome and other organizations.
The structure of patient organizations varies widely, although most operate on a voluntary basis. Even the ABG does not have its own formal structure; it is helped by its members when necessary and by demand.
The main activities of the patient organizations comprise: educating health professionals and the society, advocacy for public policies. Some are providing care for patients and support for parents. Advocacy actions include better social inclusion and education, public awareness of rare diseases, research funding and treatment for rare diseases. Most patient organizations are linked to a university, research/ reference diagnostic or treatment centre for genetic disorders.

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[237] http://www.fundaciongeiser.org/?lang=en (accessed April 23, 2013)
[238] Liljesthrom M, Armando R, 2009. Las enfermedades poco frecuentes en la Argentina: un enfoque socio-sanitario, jurídico y de investigación científica. Propuestas comunicacionales y de políticas para reducir la vulnerabilidad. (Rare diseases in Argentina: an approach from social, health, legal and research viewpoints). Report presented to the Commission of Health Polilcy of the Argentine Congress. Available at: www.fundacionfop.org.ar
[239] http://www.abram.org.br/ (accessed April 23, 2013)
[240] http://www.abg.org.br/abg/abg.asp (accessed April 23, 2013)
[241] http://fbasd.blogspot.com (accessed April 23, 2013)

China: Currently, there exists no independent patient organization officially recognized by the government. All patient organizations are registered under the administration of the National Bureau of Public Organization, Chinese Ministry of Civil Affairs. The organizations generally have an elected committee. Members include parents, family members, affected individuals and other interested people. Some groups (such as the Autism group) have a medical advisory board and few of them have a fund-raiser.

The objectives of most of the patient groups are similar and aim at seeking political legislation/policy support, raising awareness about the condition they represent in the community; offering support, advice and literature for affected individuals and their families.

Some policy and lobbying activities take place, such as making contact with the Central MoH and the Ministry of Education regarding promotion on special education for specific disorders (e.g. Down syndrome) and to the Communist Party's Central Secretary Committee and printing brochures on specific disorders.

Egypt: There are approximately 10 NGOs active in the field of genetics. They play a crucial role in supporting the community genetic counselling programme. In addition, there are 120 NGOs working in the field of maternal and child health and disabilities.

Most NGOs provide support for genetic patients. Some are funding training courses in early detection of genetic and congenital disorders for nurses and social workers and practical training in genetics for physicians. Some offer CBR services, early stimulation, behavioural modification speech therapy and other rehabilitation services to the patients at no cost.

India: Patient organizations have a long tradition in ***India***. The current number of parent organizations for genetic disorders is unknown.

There are nearly 50 haemoglobinopathy patient organizations. Some of these organizations are very active such as: *Thalassemics India, National Thalassemia Welfare Society, Thalassemic Society of India* and the *Thalassemia and Sickle Cell Society of India*. Parent organizations have formed the *"Federation of Indian Thalassemics"*.[242] The federation has created a network to coordinate services and to pressure the government to provide free treatment such as factor VIII. (Factor VIII treatment is now provided free in government hospitals in many states.)

[242] Sachdeva, A (ed.): Haemoglobinopathies. Jaypee Brothers Medical Publishers (JPB), New Delhi, 2005.

A large number of organizations is working in the field of mental retardation, cerebral palsy and autism. The organizations for mental retardation and cerebral palsy have succeeded in having a count of the disabled people in ***India*** in the national census for 2011. They have also successfully lobbied the government to provide homes for the care of the mentally handicapped.

The *Federation of Indian Thalassemics* succeeded in having the excise and custom duties on iron chelators to be reduced substantially, resulting in the availability of cheaper drugs in ***India***.
Patient organizations have been successful in having Indian Railways to provide free transport for affected persons.

5-6 organizations are in the field of muscular dystrophies. There is no single umbrella alliance under which patient organizations operate.

Most organizations organize regular meetings and CMEs on proper management and often with the help of foreign experts.

Oman has several parent/patient organizations, mainly the *Oman Society for Handicapped, Oman Association for Handicapped Children* funded by the Ministry of Social Development, the *Oman Blind & Deaf Society*, the *Early Intervention Society*, and the *Sickle Cell &Thalassemia Association*.
Their main functions include: providing funds and supervision of schools, manufacturing and distribution of hearing aids, offering educational, counselling and joint social activities, providing wheal-chairs and jobs in sheltered workshops, providing rehabilitation and vocational training, professional training, providing early intervention for children with developmental delay at the age of 1-6 and offering assistance to affected families, including medications.

The *"National Programme for Prevention of Genetic Blood Disorders"* education campaign includes lectures, media appearances, printed educational materials, advocacy sessions tailored to the needs of affected families.

The Philippines: The main parent/patient organizations are: the *Down Syndrome Association of the Philippines, Inc (DSAPI), the Philippine Society of Orphan Disorders (PSOD)* and *Balikatang Thalassemia*.

The DSAPI was established in 1992 to offer support to families who have a child with Down syndrome and to initiate, develop, promote, encourage and support programmes and projects concerning Down syndrome.

Through the efforts of the parents, the month of February was declared as the *"National Down Syndrome Consciousness Month"* by the President of the Philippines

in 2002.[243] Based on this proclamation government departments (DoH, Department of Education, Department of Labor and Employment, Department of Social Welfare and Development) and related agencies and appropriate NGOs were enjoined to support and cooperate with the activities of the DSAPI.[244]

The PSOD was founded in June 2006 with a main objective to continue the efforts of physicians (mostly from the IHG-NIH-UP) to ensure sustainability of medical and financial support of patients with "rare disorders."

The PSOD is currently lobbying the enactment of the *"Rare Disease Act of the Philippines."*[245]

Project Rare Program was launched by the PSOD in 2009. The project kicked off a public awareness campaign to support the care of children born and afflicted with rare diseases. A series of activities aim at (i) increasing the registry of patients and referring them to the IHG-NIH for diagnosis and available treatment, (ii) building a network of voluntary partners and friends and, (iii) building an endowment fund to sustain the lifelong medical treatment and therapies of patient members.

The endowment fund helps to sustain the lifelong treatments of patients as well as to provide financial support for research on rare disorders. The fund also aims at making the PSOD self-sustaining. An emergency fund helps to address immediate needs of patients with medical emergencies. This can include, but is not limited to, providing life-saving medicines, hospitalization, purchase of supportive medical devices, and other support.

The *Balikatang Thalassemia* foundation was founded in 1995 with the main objective to provide medical assistance, education, and counselling to thalassemic patients, their parents, and families. To make thalassemia a public health concern and to ensure government support for its different programmes, the organization supported the enactment of the *National Services Act of 1994* which was activated in 1999 as the Republic Act 7719. This act mandated the Children's Medical Center Philippines

[243] Proclamation No. 157, February 2002

[244] One of the highlights of the "Down Syndrome Consciousness Month" is the Happy Walk. In 2010, over 3000 children/adults with Down syndrome, parents, volunteers and other supporters like doctors, therapists, teachers, students from different colleges and universities, TV, Movie and sports personalities, and other interested parties participated. This activity was simultaneously conducted in Manila (Luzon), Cebu (Visayas) and Davao (Mindanao) to promote Down syndrome awareness nationwide.

[245] Senate Bill No. 3087 (http://www.senate.gov.ph/lisdata/103909260!.pdf, accessed May 16, 2013) was introduced February, 2009, and the congress introduced House Bill 6937 October, 2009.
The bill seeks to establish a system that will ensure the early diagnosis and treatment of rare diseases. The bill provides the creation of a rare disease programme at the National Department of Health. The programme seeks to ensure the provision of early and sustainable care for patients suffering from rare diseases, supervise the implementation of a research programme on rare diseases, and coordinate current activities of the National Department of Health to provide patients with rare diseases and their families with access to adequate medical care, health information, and healthcare products. The bill will support public education and information campaigns on rare diseases, health professional training, and establish a system to coordinate a research & development initiatives and resource generation efforts among relevant agencies of government and the private sector to improve the quality of life of patients with rare diseases and their families.

(now the Dr. Fe Del Mundo Medical Center) as the "Thalassemia Center of the Philippines".

The National Council for Disability Affairs (NCDA) [formerly National Council for the Welfare of Disabled Persons (NCWDP)] is the national government agency mandated to formulate policies and coordinate the activities of all agencies, whether public or private, concerning disability issues and concerns.[246]

The NCDA is the lead agency tasked to steer the course of programme development for persons with disabilities and the delivery of services to the sector. The NCDA monitors the implementation of laws to ensure the protection of persons with disabilities' (PWD) civil and political rights.

South Africa: Parent/patient groups for a number of the common genetic conditions are established. Many were started in association with and with encouragement from the *Southern African Inherited Disorders Association (SAIDA)*. This association was initiated by a medical geneticist and genetic counsellor in 1975, in response to the request of a couple with a child with Tay Sachs disease. There are currently 25 groups who are members of SAIDA, a further 24 groups who have been members in the past, and a few who are loosely connected or function independently. Most groups operate from the big cities and offer country-wide services to anyone affected by or interested in their specific disorder.

Some policy and lobbying activities take place, such as making contact with the National DoH Genetic Services division to print brochures on specific disorders (e.g. Turners syndrome), or the Department of Education regarding inclusion and mainstreaming policies (e.g. for children with Down syndrome), or the Department of Labour regarding employment for disabled people. Advocacy activities also occur (e.g. self-advocacy courses for adults with Down syndrome), and empowerment activities (e.g. affected individuals seeking free provision of sun-barrier cream for people with albinism at government hospitals).

[246] http://www.ncda.gov.ph (accessed April 30, 2013)

Below **Box 8.1** lists the most important parent/patient organizations in GenTEE countries.

Box 8.1 Parent & Patient Organizations in GenTEE countries (2012)

Argentina	Asociacion Sindrome de Down de la Republica Argentina (ASDRA) (http://www.asdra.org.ar) Geiser Foundation (http://www.fundaciongeiser.org/?lang=en)
Brazil	Aliança Brasileira de Genetica (http://www.abg.org.br) Associação Brasileira de Assistência a Mucoviscidose (http://www.abram.org.br) Federação Brasileira das Associações Síndrome de Down (http://www.federacaodown.org.br)
China	Home for Premature Babies
Egypt	Egyptian Thalassaemic Friends Association
India	Down's Syndrome Care Association (DSCA) (http://www.dscaindia.org) Down Syndrome Federation of India (http://www.downsyndrome.in) Indian Association of Muscular Dystrophy (http://www.iamd.in) Muscular Dystrophy Foundation India (http://www.mdfindia.org/stage/index.php) Thalassemics India (http://www.thalassemicsindia.org) National Thalassemia Welfare Society (http://www.thalassemiaindia.org) Thalassemia and Sickle Cell Society of India (http://www.tscs.in) Muskaan (Parents organization for children with mental retardation (http://muskaan-delhi.com/) Action for Autism (http://www.autism-india.org/) Fragile X syndrome society of India (http://www.fragilex.in)
Oman	Early Intervention Society for Children with Disabilities (http://www.aei.org.om) Oman Blind & Deaf Society Oman Hereditary Blood Order Association (http://www.omancares.org/en) Oman Society for Handicapped
The Philippines	Balikatang Thalassemia (http://www.thalassemiapatientsandfriends.com/) Down Syndrome Association of the Philippines, Inc (DSAPI) (http://dsapi.org) Philippine Society of Orphan Disorders (PSOD) (http://www.psod.org.ph/ver2/)
South Africa	Down Syndrome South Africa (DSSA) (http://www.downsyndrome.org.za) Southern African Inherited Disorders Association (SAIDA) (http://www.saida.org.za/)

Funding

Argentina
Patient organizations get funding from their own members, from private foundations and from the government

Brazil
Most patient organizations are financed by donations and by their members; support from the society in general is quite rare. There is no information regarding financing of the organizations.

China
Fund raising is generally undertaken in a very limited way, unless the association is well recognized and a financial professional is involved in the actual operation/organization of the association. Most support groups receive very little funding via various sources. Government seldom funds the small public organizations, the majority of the funding come from membership fees and donations. Some large associations may own their own retailing or publishing company as their continuous solid source of funding.

Egypt
Sources of funding are donations through fundraising activities.

India
Patient organizations are funded by donations, memberships dues and are sometimes supported by the government.

Oman
Sources of funding are donations through fundraising activities

The Philippines
DSAPI is a non-stock, non-profit organization, whose members are volunteers. The primary sources of funding come from membership fees, voluntary contributions and donations from friends and supporters.

PSOD is a non-stock, non-profit organization, the endowment fund and the emergency fund sustain the lifelong medical treatment and therapies of patient members.

Balikatang Thalassemia is a non-stock, non-profit, non-political corporation founded in 1995.

South Africa

Fund raising is generally undertaken in a limited way, unless a fund-raiser is employed or a member of the group undertakes this responsibility systematically (e.g. *Retina South Africa*, who have a large budget and support research). Most support groups receive very little or no regular government funding (with the exception of the *Cleft Pals Support Group* that has some government funding) and fund themselves through membership fees and donations. Where fund-raising is undertaken, most funding comes from private businesses, corporate and individual donors, sometimes from pharmaceutical companies, and in a few cases from international bodies (e.g. the *Haemophilia Foundation* receives some support from the *World Federation for Haemophilia*) and grants from the national lottery.

Public education in genetics

Argentina

Primary prevention measures focus on folic acid fortification, rubella immunization, campaigns against tobacco and alcohol

Brazil

There is no specific focus from the government or the National MoH on the prevention of congenital / genetic disorders.

Brazil has a policy of flour fortification (wheat and maize); salt is iodised.

Rubella immunization is available and the programme includes specific awareness campaigns for women in reproductive age;

There are labels in alcoholic beverages and cigarettes informing about consumption risks during pregnancy;

Women are informed about the importance of newborn screening during prenatal care.

China

Due to the increased awareness of the central government, protocols have been established to address congenital/genetic disorders. ***China*** provides free folic acid supplement to women from 3 months before to 3 months after the beginning of pregnancy. As a result the prevalence of neural tube defects has fallen by approx. 30% nationally, and by 50% [247] in some provinces that had higher prevalence rates of neural tube defects previously. Salt has been iodised for many years.

Lay groups, such as labour unions, women's unions or residential societies, may routinely organize talks given by human genetics professionals. Although genetic counselling is not recognized as a health profession currently in ***China***, obstetricians and gynaecologists are counselling and giving educational lectures to both professionals and to the public. Maternal & child health hospitals and other professional/governmental groups (including voluntary groups such as *Best Baby Association* and *National Health and Family Planning Commission*[248]) may compile leaflets for distribution on a number of common disorders, as well as on PND and genetic counselling services.

Egypt

The MOH&P has adopted several ways to improve the knowledge, attitude and practice of the women in the childbearing period for the prevention of congenital and genetic disorders including health education seminars, social mobilization

[247] Data provided by Nanbert Zhong, Peking University Center of Medical Genetics, Beijing, People's Republic of China.

[248] http://www.npfpc.gov.cn/ (accessed April 25, 2013)

campaigns, designing educational leaflets and brochures for proper nutrition, safe pregnancy and safe motherhood. A yearly budget is allocated for training and education of nurses and community outreach visitors on health education including the proper methods to deliver health education massages for the community and certain specific messages for the preventive national programmes like the importance of antenatal visits, inter-conceptual care programme and community genetic counselling programme.

India

The Indian government focuses on the reduction of the prevalence of low birth weight and premature deliveries.
The use of folic acid before pregnancy is advocated by health professionals.
The *Micronutrient Initiative India*[249] supports India's salt iodization programme and has initiated pilot projects on iron folic acid.

Oman

A number of educational materials has been produced in the past 10 years including management guidelines and educational booklets[250]:
Education is provided through media including radio and TV interviews, TV spots and mobile telephone messages. Education activities in schools and colleges include lectures, public events and a marathon.

The *National Programme for the Control of Genetic Blood Disorders* provides community counselling and education for haemoglobinopathies.

The Philippines

There is a national campaign for the newborn screening panel including since 2012 six disorders (CH, CAH, galactosemia, G6PD deficiency, PKU and Maple Syrup Urine Disease (MSUD)).

The *Volunteer Youth Leaders for Health - Philippines* (VYLH), a network of youth leader volunteers from the different youth organizations, assists in the advocacy of the campaign on folic acid awareness. Currently, the network is doing advocacy and promotional work in their respective schools and communities on increasing

[249] http://www.micronutrient.org/english/view.asp?x=603 (accessed May 8, 2013)
[250] Published MoH materials include:
1."Facts of Life" for school and university students explaining healthy living (has genetic disease section),
2. Picture guide "Understanding heredity" for individuals with limited ability to read explaining heredity with help of visual graphics (units of heredity, chromosomal inheritance and basis of Autosomal Recessive inheritance.),
3. Information for: Sickle Cell disease patients with explanation of pathology, possible treatments and inheritance
4. Information for Beta-Thalassemia patients with explanation of pathology, possible treatments and inheritance
5. Information for understanding of pathology of G6PD deficiency, avoidance of crises and inheritance
6. Information for understanding Down syndrome with medical management plan and rehabilitation
7. Leaflets produced: Carriers of Sickle Cell, Carriers of Beta-Thalassemia, G6PD Deficient, Explaining x-linked inheritance

awareness among women in their reproductive age on the significance of folic acid supplementation. Also included in the activities are: increasing public awareness in saving babies from mental retardation and death through newborn screening and lobbying public support for the passage of the *Rare Disease Act*. The youth volunteers are handing out promotional flyers and posters on newborn screening; delivering lectures and exhibits on folic acid; organizing and conducting symposia on newborn screening, congenital and genetic disorders surveillance and folic acid campaign; and conducting signature campaign for the *Rare Disease Act*.

South Africa

Due to concerted lobbying, ***South Africa*** has had fortification of basic foods, such as bread and maize meal, for some years, and as a result the prevalence of neural tube defects has fallen by 30%.[251] Salt has been iodised for many years preventing most cases of postnatal iodine deficiency disorders and goitre.

Public and professional education, covering recognition and prevention of congenital disorders, takes place at many levels. Health professionals, including nurses, receive some basic teaching in medical genetics during their degree and diploma studies, as well as in-service training when they are employed. Lay public groups, such as rotary clubs and women's groups, have talks from human genetics professionals when they request them. These professionals also give radio interviews when asked to do so. Genetic counsellors give educational lectures to a number of professional and lay groups[252] and compile leaflets for distribution on a number of common disorders (e.g. genetics of breast cancer), as well as on PND and genetic counselling services. SAIDA puts out an annual educational newsletter, which is distributed to professional and lay groups.

Furthermore, SAIDA, with support funding from the Human Genetics sub-directorate, National DoH, and more recently the national lottery, offers a short-term course, the *Medical Genetic Education Programme* (MGEP) on basic genetics for primary healthcare workers. The course is directed at doctors and nurses working in primary healthcare clinics and covers aspects of basic genetics, including the identification, basic treatment and counselling (particularly breaking bad news) for affected children and their parents. The course consists of a combination of contact days, with lectures and workshops, as well as self-directed learning by using a provided text-book. In this manner, it is hoped that more patients and their families are reached and that earlier diagnoses can be made, as well as earlier intervention and care implemented. Nurses and doctors alike are participating in these courses, and it is hoped that a

[251] Sayed A-R, Bourne D, Pattinson R, Nixon J, Henderson B (2008). Decline in the prevalence of neural tube defects following folic acid fortification and its cost-benefit in South Africa. Birth Defects Research (Part A); 82: 211-216.

[252] Kromberg JGR, Krause A, Wessels T (2009). Roles of Genetic Counsellors in South Africa. SA Society for Human Genetics 13th congress, 5-8 April 2009, Stellenbosch, South Africa. Book of Abstracts p36.

result will be more accurate reporting of congenital and genetic disorders, which will add to the epidemiological data for the country.

One day workshops are held, occasionally, for general practitioners in Johannesburg, and in some other urban areas of the country. However, due to the inadequate number of medical geneticists and genetic counsellors working in the country, availability of skilled human resources for the expansion of community education programmes remains a challenge.

Civil society engagement: Patient organisations in GenTEE countries

Comment by Alastair Kent, Genetic Alliance UK

Although each of the GenTEE countries are different in terms of their social structures and their health care systems they all have emerging patient support organisations. While there are clear differences between each of the participant countries in the extent to which these groups have emerged, there are a number of similarities that can be identified.

The first of these, and one of the most significant, is the hand to mouth existence forced on many patient organisations by the lack of any infra-structural funding from the state. Whilst no-one has a right to financial support simply by virtue of existing, the absence of reliable core funding forces patient groups back on their own resources supplemented by what can be raised through fundraising and other efforts. For groups which comprise patients and carers, already constrained by the limitations of their condition and the impact this has on their everyday opportunities, this can be a significant limitation to the achievement of their potential.

A second common element is the way in which families affected by relatively common genetic conditions tend to be the ones that come together to form support groups first. Thus where haemoglobin disorders are common there tends to be a better established group than for many of the rarer conditions. Down syndrome too is an early condition for the emergence of support groups.

Thirdly, coverage is by no means universal. Many patients do not have a support group to turn to for help, and where these exist it is perhaps as a result of the initiative of a particularly charismatic individual, possibly working in partnership with a clinician of an established centre.

A fourth feature groups have in common is that, despite any progress they might make in improving services and support for their member families from the statutory sector, direct services and support, particularly the dissemination of practical advice on day to day issues remains a key strand of their work.

There is an emerging trend for condition specific support groups to come together in alliances to create common ground and generate the critical mass necessary to be effective in the strategic advocacy role that many aspire to. This is most notable in ***South Africa*** and ***Brazil***, where established alliances have played a significant role in profile raising and the generation of awareness, but there are signs of this trend elsewhere too.

Looking at the GenTEE country reports as a whole, what can be observed is a growing recognition of the legitimacy of the patient and family perspective (albeit to a varying degree for instance there are still strict limitations for establishing parent and patient organizations in ***China*** and ***Oman***) on what and how services and support can and should be provided.

This is developing as a collaborative issue, with the different contributions from all; key stakeholders gradually being recognised and made a formal element of the process whereby clinical and medical genetics services are provided to communities and populations. This has to be a welcome development, but there are major challenges to be overcome before the patient and family voice is automatically seen as part of the process for determining service provision and for the promotion of the opportunity for high quality research into causes and cure for genetic disease. The grass roots movement of the patient community is, on the basis of these country reports at least, determined to play a role in shaping the future of genetic medicine and genetic services, and the organisations representing patients and families have a vital role to play in bringing this about. To succeed they will need support, resources and most of all the opportunity to be at those tables where decisions are taken as a right, not as a favour. This will necessitate the other stakeholders moving over to make space for this new entrant. It will require systems and established ways of working to be adjusted to allow this voice to be heard, and it will require investment in promoting and developing the voice of patient organisations so they can articulate the needs and issues of their members in ways that can be heard and responded to. The evidence of this survey is that there are encouraging steps being taken towards this goal. This is welcome, but the development is fragile in many situations and needs to be nurtured to full development. This is the task for the coming decade.

IX Drivers and barriers for genetic services development

Argentina:

Drivers:

Policies to improve capacity and access to services

In ***Argentina***, the National MoH has recently shown interest in improving access to and performance of the clinical genetics services throughout the country.

A *National Commission on Genetics and Health*, appointed in 2005, conducted countrywide studies on the situation of genetic services and submitted a proposal to the Ministry for their improvement. The proposal asked that the number of positions of clinical geneticists in public hospitals be increased and that the laboratory equipment for cytogenetics and molecular genetics in several hospitals countrywide be modernized and expanded. The Commission conducted a survey of genetic services and proposed the organization of a network of genetic services to maximize their efficiency, avoid duplication of services, and channel referrals in a regionalized manner.[253]

Acting on such recommendations, in 2010 the National MoH started some actions to improve the capacity of the existing genetic units in the public system.

In addition, RENAC has begun in 2009 in selected provinces, centrally coordinated by the National Medical Genetics Center, an agency of the National MoH. In the period 2009-2011, 182,070 live neonates (28% of the total annual number of births of the country) were examined in 107 hospitals, finding 3,234 neonates with major structural defects (1.78%).[254] This will lead to the availability of better actionable data for informed policy decision making to improve services.

Service providers' initiatives

The National Pediatric Hospital Garrahan has taken a leadership role in conducting training in clinical genetics for primary care health personnel in several underserved areas of the country.

[253] Alba A, Barbero P, Barreiro C, Chertkoff L, Dain L, Ferreiro V, Francipane L, Frechtel G, Gallego M, Liascovich R, Meroni ME, Rozental S (2007). Diseño y organización de una Red Nacional de Genética Médica (Design and organization of a national network of medical genetics.) Spanish. Unpublished, available from the author.
[254] RENAC-Ar (2012). Registro Nacional de Anomaías Congénitas de Argentina. Publication of the Ministry of Health of Argentina..

Barriers:

Barriers are mainly bureaucratic as each province has its own set of policies and budget, and the National MoH does not have much leverage or resources to impose policies.

Brazil:

Drivers:

Policies to improve capacity and access to services

The Brazilian MoH published a decree in 2009, which proposes the creation of a *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS).[255] However, the decree is still waiting for implementation. Currently, a commission is elaborating a policy for rare disorders which may include a policy for genetic disorders (expected to be implemented in 2013).

Brazil has made significant investments to fund research in medical genetics and genomics. The main funder is the public sector, both state and federal.

Barriers:

- Delayed implementation of national policies to improve genetic services;
- Geographical unavailability of services;
- Lack of universal coverage;
- Availability of services at primary care level very limited;
- Legal constraints concerning abortion;
- Medical genetics not a formal specialty in the Unified Health System (SUS) leading to few available job positions for geneticists.

Future outlook:

Organizing a network in clinical genetics

[255] Brasil (2009a), Ministério da Saúde, portaria GM no. 81, 20 de janeiro de 2009 – Institui, no ambito do Sistema Único de Saude (SUS), a Politica Nacional de Atenção Integral em Genetica Clinica. Diário Oficial da União 21/01/2009

The network configuration of services in clinical genetics (regionalized, hierarchical and functional, as recommended in the creation of the SUS) will be a crucial item for care in medical genetics.

Government initiatives that already exist need to be consolidated

In addition to formalizing and carrying out the organization of a network in clinical genetics in ***Brazil***, other actions need to be implemented for the system not only to properly function, but also to be gradually expanded and adapted to the country´s growing needs. Government initiatives that already exist need to be consolidated, and non-governmental programmes may eventually be added and enrich the system. As examples of optimization and integration, informing city officials about the importance of the correct completion of "Field 34" of the “Liveborn Declaration” (“Field 34” addresses congenital anomalies present at birth) needs to be encouraged.[256]

Integration of genetic services into the Unified Health System/SUS

Considering the magnitude of the impact that congenital and genetic disorders already have on health, in a country like ***Brazil***, as well as all the perspectives generated by the advances in this field, it must be assured that genetic services and testing are appropriately integrated into health care in Brazil and become part of the SUS.

Supporting parent and patient organizations

The importance of patient-parent organizations should be reinforced; besides offering support and comfort to their members, such associations have as objectives the dissemination of information among lay people and to physicians. These non-governmental associations can play a fundamental role of introducing new topics on the political agenda.

Improvement of education in genetics including ethical issues

The issue of prevention needs to be addressed including MToP.

China:

Drivers:

Policies to improve capacity and access to services

[256] Former “Field 34”has been now divided in two different field in the liveborn declaration.

China is a fast developing country with the largest population size in the world. The economic situation and quality of life of the population have improved dramatically in the past 20 years. Since the founding of the Republic in 1949, the central government has given a great deal of attention to maternal and child health. As a consequence, during the last two decades, ***China*** made substantial progress in reducing maternal, infant and under-five mortality.

Barriers:

Nevertheless, national figures for maternal, infant health indicators mask large disparities, which exist between urban and rural populations, and across different regions of ***China***. There is limited access to medical genetic services and maternal and infant death rates are highest among the rural poor and migrant population, and in those regions with least access to antenatal and intrapartum care, such as the western provinces.

Future outlook:

The governmental awareness in health service is moving towards coverage for the whole population as well as to the development of state of the art molecular diagnostic centers. It is widely believed that in the short future, the imbalance of health service between urban and rural areas will be improved, and more molecular tests will be available for the public.

Egypt[257]

India:

Drivers:

The control of infectious diseases through immunizations and therapy has led to the emergence of congenital and genetic disorders as important causes of morbidity and mortality in urban areas. The government has realized the burden of non-communicable disorders. Parent and patient organizations have put pressure on the government to pay attention to the burden of congenital and genetic disorders like autism, haemophilia and thalassemia. States now provide free treatment to patients with thalassemia and provide free factor VIII therapy to patients with haemophilia through government hospitals. Recently the government has launched an ambitious programme to screen for congenital disorders such as blindness, deafness, cleft lip and palate, autism, cognitive decline and others, starting in about 20 districts and then spreading this programme to all districts in ***India***.[258] The *National Rural Health*

[257] Not addressed in the survey.
[258] Press Information Bureau, Government of India, 27 March, 2013. Online available at http://pib.nic.in/newsite/erelease.aspx?relid=92045 (accessed April 12, 2013)

Mission (NRHM)[259] established by the government of ***India*** is improving primary care for easily recognizable congenital defects.

Funding agencies (DST, DBT, ICMR, CSIR) are investing heavily in genetic biotechnologies and genetic research realizing the potential of the new genetic/genomic technologies available in the post-human genome project era.

Barriers:

The sheer size of the country and its vast population, living predominantly in the rural areas, has hindered the provision of genetic services to many people. The cost of genetic testing services, the lack of medical geneticists and genetic counsellors is also a barrier. The mindset of the government administrators (priority given to infectious disorders, limited understanding of the burden of congenital and genetic disorders and of the scope of available interventions for care and prevention) is not yet attuned to providing genetic services and therefore administrators still lack political will and commitment to these services. The money is there but genetics still has a very low priority for the government.

Oman:

Drivers:

The main drivers for the development of medical genetic services are

(i) the increasing recognition by policy-makers of community needs for genetic services and
(ii) the increasing availability of new genetic information evolving from the advancement of the science of genetics and better understanding of genetic predisposition to adult-onset disorders.

Policies to improve capacity and access to services

The MoH Health of The Sultanate of ***Oman*** has recognized the need for genetic services and genetic technologies as means for controlling genetic diseases in the Sultanate and aims at ensuring high standard medical care in the era of rapidly expanding genetic science and biotechnology.

[259] http://www.mohfw.nic.in/NRHM.htm (accessed April 17, 2013)

A *National Committee for the Prevention of Genetic Diseases* was established in 2004 and includes representatives from Ministries of Health, Education, Social Affairs, Information and National Economy.

In 2005, the MoH of the Sultanate of ***Oman*** published its 7th *Five-Year Plan for Health Development* (2006-2010)[260] which included a national strategic plan on genetic diseases in order to reduce the infant and under-five morbidity and mortality to the lowest international rates.

Among the strategies implemented to achieve these objectives, a National Genetic Centre to provide clinical and laboratory diagnostic services was established to provide care services and to support community prevention programmes, conducting training activities and research in the field of genetic health. The construction of the National Genetic Centre has been completed in December 2012.

Extensive training of Omani nationals is underway to prepare for functions of the National Genetic Centre.

Barriers:

Lack of Omani specialists trained in genetic counselling and bioinformatics. ***Oman*** still relies on "consultants" from abroad.
Limited availability of genetic testing services for late-onset disorders and rare disorders.
Due to the scarcity of skilled experts current services are ill-prepared to take advantage of the new genetics/genomics technologies and to use new genetic/genomic knowledge in clinical patient pathways.

The Philippines:

Drivers:

Policies to improve capacity and access to services

The Philippines is one of the most active countries in Southeast Asia in regard to genetic services development.

Service providers' initiatives

The IHG-NIH has taken a leading role to improve the availability and capacity of genetic services especially in regard to the successful implementation of a national

[260] Ministry of Health, Sultanate of Oman (2005) 7th Five-Year Plan for Health Development (2006-2010). Available at http://www.moh.gov.om/en/nv_menu.php?o=fiveyearplan/fiveyearPlan.htm&SP=1 (accessed 17 October, 2012)

newborn screening programme, developing counselling services and training of manpower in genetics.

Barriers:

Being a developing middle-income country, ***the Philippines*** is faced with the challenge of providing healthcare for all Filipinos. Although one of the most active countries in southeast Asia with regard to genetics, the country still has a shortage of geneticists and genetic counsellors. Difficulties exist for continued research and integration of healthcare services into the public health system. The main barriers to accessing genetic services in ***the Philippines*** are:

(i) **financial**, since most families cannot afford out-of-pocket expenses for the expensive genetic testing and treatment;
(ii) **geographical**, being an archipelago of 7,107 islands;
(iii) **lack of awareness** among different stakeholders, i.e., health professionals and parents;
(iv) **compromised access** to genetic services at the regional and provincial level; and
(v) **lack of geneticists** and **genetic counsellors**.

However, despite these shortcomings, the IHG-NIH views a promising future for medical genetics in the country, with the help of the government and support of the community.

South Africa

Drivers:

Policies to improve capacity and access to services

The Constitution, Laws, and Policy Guidelines for the Management and Prevention of Genetic Disorders released in 2001, all mandate for the development of services for the care and prevention of congenital and genetic disorders. However, as noted above, the intention of these has been lost in translation.

Service providers' initiatives

Medical genetic services was mainly developed in the country by leading medical geneticists prior to the onset of the HIV/AIDS and TB epidemics These service would have provided an excellent base on which a more comprehensive service can be built in the future.

There are four academic departments of human genetics in four of the major universities situated in three provinces which have already set up clinical services, compatible with any in developed countries, as well as laboratory services, which have the expertise to offer genetic testing services to the country and to the rest of Africa.

They have also developed a research capacity so that local genetic disorders can be investigated on many different levels.

Two of these universities, in collaboration with the NHLS, have established training for all the categories of expert staff required to run a sophisticated genetic service.

However, the aging of laboratory equipment and lack of financial support for the introduction of new technology are making it difficult for these capable scientists to keep up to date with new developments in the field.

Although expansion is difficult in the current circumstances, members of the South African human genetics community are networking with interested people in the rest of Africa and, in March 2011, the first combined congress was organized by the *SA Society of Human Genetics* (SASHG), in conjunction with the *African Society of Human Genetics* (AfrSHG), and held in Cape Town. At this meeting there were international experts, as well as those from Africa, networking and sharing their expertise, knowledge, insights and needs, which will both benefit and stimulate the field of human genetics on the African continent in future. At this stage there is some hope that the investment by international and local agencies in, for example, the

Southern African Human Genome[261] project and the *Human Heredity and Health in Africa* (H3Africa)[262] programme, may raise the profile of medical genetic services and contribute to their upgrading.

Barriers:

Burden of HIV infection and TB epidemics

South Africa is a country in transition. However, the proportion of the global burden of disease borne by ***South Africa*** with a population of only 49 million is disproportionately high. The total disability adjusted life years for high burden diseases in ***South Africa*** is almost equivalent to that of Bangladesh, which has a population three times as large and living in much worse poverty.[263] One of the greatest challenges it faces is the control of the concomitant HIV and TB epidemics. In 2007, the country, with 0.7% of the world's population, had 17% of the global burden of HIV infection, and one of the world's worst TB epidemics, compounded by rising drug resistance and HIV co-infection.[264] ***South Africa*** is currently underperforming in its efforts to control HIV and, although it has the resources and capability to rise to these challenges, it has not been able to deliver on the four priorities listed in the Strategic Plan for ***South Africa*** for HIV/AIDS.[265] Since medical genetics services are, presently, being developed and delivered against this background, it is not surprising that the recommendations in the *Policy Guidelines for the Management and Prevention of Genetic Disorders*[266] have not been met.

Declining political commitment to invest in the provision of clinical genetic services

The commitment to provide clinical genetic services in ***South Africa*** has declined over the last few years. There is continued discussion as to whether these services should be centrally co-ordinated from the National DoH and provided by the provincial Departments of Health, as occurs in the Western Cape and Free State, or whether they should be all be provided by the NHLS, as occurs in Gauteng. Until this issue is resolved or another solution found, clinical genetics services will not improve and are at risk of deterioration. Laboratory genetic services are also at risk from increasing demand for tests in the face of reducing staff availability.

[261] http://www.pub.ac.za/index.php?option=com_content&view=article&id=66&Itemid=131 (accessed April 17, 2013)
[262] http://h3africa.org/ (accessed April 17, 2013)
[263] Lancet, paper 6, 2009. In: Lancet Series (2009) Health in South Africa 1–6. Online available at http://www.thelancet.com/series/health-in-south-africa (accessed May 16, 2013).
[264] Lancet, paper 3 abstract, 2009. In: Lancet Series (2009) Health in South Africa 1–6. Online available at http://www.thelancet.com/series/health-in-south-africa (accessed May 16, 2013).
[265] Lancet, paper 3, 2009. In: Lancet Series (2009) Health in South Africa 1–6. Online available at http://www.thelancet.com/series/health-in-south-africa (accessed May 16, 2013).
[266] Department of Health, South Africa (2001). Policy Guidelines for the management and prevention of Genetic Disorders, Birth Defects and Disabilities. Department of Health, Pretoria, South Africa.

These issues will be considered by the newly appointed *NHLS Expert Committee for Medical Genetics Services*, and it is hoped that solutions to the situation will be found.

Lack of employment opportunities for qualified professionals leading to brain drain and understaffed services

This has become a major problem in the last two years. Two medical geneticists have recently qualified and at least another four will qualify in the next two years. No posts are available for them and are unlikely to become available in the foreseeable future. One newly qualified medical geneticist has already left the country. A similar situation has developed for genetic counsellors.

Medical genetic laboratory service units and staffing is also being reduced resulting in decreasing opportunities for medical genetic diagnostic scientists and technologists.

Inequity of access leading to inaccessibility of genetic services for the poorer population

The reducing available clinical genetic services are largely available in three centres, Cape Town, Johannesburg and Bloemfontein. Geographic (distance) and expense therefore act a serious barriers to most of the public accessing genetic services.

Future outlook:

Development of medical genetic services, in the near future, will depend partly on increasing the awareness of genetic disorders, partly on lobbying the decision-makers in the health departments and NHLS, partly on the control and lessening of the HIV/AIDS epidemic, and mostly on the provision of more employment opportunities for qualified professionals. At present, the available staff can only meet at most 10% of the country's genetics needs (based on a rough calculation of the genetic burden of disease). Further technological development (together with purchase of the necessary laboratory equipment) should be planned for, so that South Africa can approach the level of developed countries. Both political will and financial commitment are required to move this enterprise forward. Pressure is being brought to bear on key members of the National DoH and its provincial subsidiaries by medical genetics professionals, as well as by genetic support group representatives, to respond to the basic genetic needs of ***South Africa*** and make an adequate and appropriate medical genetic service available. Given current circumstances these are unlikely to succeed in the foreseeable future.

Drivers and barriers for genetic services development

The GenTEE survey provides a detailed overview over the current state of genetic service and testing provision in the participating countries and addresses the challenges these countries face to develop an equitable service infrastructure. As of today in most countries (except ***Oman*** where universal coverage facilitates the access to services) **genetic services are mainly accessible for the affluent urban upper-middle and upper classes** who can afford to pay out-of-pocket for services in the private sector.

Lack of health professionals educated in genetics and health workforce training in genetics is a ubiquitous problem in all GenTEE countries.

In countries like ***Brazil***, ***China***, ***India*** and ***South Africa*** that invest heavily in genomic science and research, **there is a striking mismatch between highly developed research capacities and the non-availability of equitable services** that are prepared to take advantage of genetic/genomic technologies and information in order to improve the care for their patients.

Nevertheless – maybe with the exception of ***South Africa*** – in all GenTEE countries **positive developments to improve genetic service structures can be observed** – although in some countries developments can be painstakingly slow. The major challenges clearly lie in providing equitable services and integrating genetics and genomics into existing public health care services.

The message is clear: **in order to reap the potential benefits that the rapid development of genetic/genomic technologies and knowledge brings, the current service infrastructure needs to be strengthened in all GenTEE countries**. This should ensure the successful translation of genetic/genomic laboratory and academic research into quality assured pathways and the improvement of both the individual patient outcomes and the overall population health.

Below a short SWOT (Strengths, Weaknesses, Opportunities and Threats) overview on the current state of genetic services is provided.

SWOT analysis genetic testing services in GenTEE countries

Country	Strengths	Weaknesses	Opportunities	Threats
Argentina 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • participates in the ongoing ECLAMC • started RENAC	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact on congenital and genetic disorders on health services • no data available on the prevalence of hereditary "late-onset disorders"	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • RENAC started in 2009 in selected provinces and its expansion is ongoing;. current coverage: 50% of births in the public system, 28% of births in the private system.	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease burden • without effective interventions in place in the public health care sector, the projected number of infants born with serious congenital and genetic disorders will increase over the next decades with important service implications • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-informed decisions on planned service development

Country	Strengths	Weaknesses	Opportunities	Threats
Argentina	**Availability of key genetic services:** • mandatory newborn screening programmes available (covering 10 disorders and two thirds of the population, close to 100% in the city of Buenos Aires) • genetic counselling services established in over 40 genetic clinics in the public health care system • preconception care services available	**Availability of key genetic services:** • the development of services in the private sector is opportunistic and mostly market-driven • genetic screening tests (except for newborn screening) are not widely available • carrier testing not available • reproductive genetic services and MToP not available in the public domain due to the illegality of abortion	**Availability of key genetic services:** • acceptable number of genetic units and genetic testing services, however primarily at tertiary care level • increasing numbers of primary care centers with health workers trained to detect and refer genetic disorders • increasing numbers of public hospitals in the provinces with telemedicine in place for genetic consultation with tertiary centers	**Availability of key genetic services:** • the legal restriction of abortion prevents the development of PND, PGD and MToP services in the public domain
	Access to genetic services: • there is an extended network of hospitals and health centers in the public sector, providing a basis for the expansion and improvement of access and quality of genetic services	**Access to genetic services:** • inequitable access to services, many services are located in the private sector and have to be paid for out-of-pocket and are located only in urban areas • lack of universal health coverage; social and private insurances usually deny coverage of genetic services on the grounds of pre-existing condition • coverage of the national newborn screening programmes less than adequate, given it is mandatory by law. • lack of expertise and skill gaps in recognizing congenital and genetic disorders by primary care	**Access to genetic services:** • genetic centres in tertiary care hospitals run telemedicine programmes for genetic consultations to overcome geographical barriers • the private for profit health sector is under increased public scrutiny for the barriers imposed to patients with genetic disorders, and legislative action to enforce equitable services is in course; • health professionals are increasingly interested in medical genetics	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • mostly the affluent urban upper-middle and upper classes can afford services • excessive waiting lists in public health sector genetic services that implicitly lead to non-transparent prioritization and rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps to recognize congenital and genetic

Country	Strengths	Weaknesses	Opportunities	Threats
Argentina 		providers impedes the route through needed diagnostic care and prevention services		disorders result in delayed referral
	Current state of genetic services: • recognition of medical genetics as a medical specialty • professionalization through the establishment of professional bodies and scientific societies' development of qualification standards • postgraduate programmes for laboratory services available • education programmes in medical genetics and genetic counselling available for non-genetic health professionals • the basic testing technologies required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and DNA testing are available in major centres • pharmacogenetic testing being introduced selectively • public genetic services are part of a national network of 1,319 public hospitals and 6,290 PHC centres	**Current state of genetic services:** • the development of services in the private sector is opportunistic and mostly technology and market driven • genetics knowledge of physicians is poor as most medical schools do not include meaningful teaching in genetics in their curricula • underfunded and understaffed public health sector services that are unable to deliver the volume of needed services are the norm • unsatisfactory referral structures in the public domain, all too often patients affected with genetic conditions must find their own way to a tertiary hospital to find genetic services • there are no official agencies that control or monitor the analytical validity of tests, quality assessment of laboratory results relies mostly on the voluntary decision of the laboratory directors to participate in a quality control programme,	**Current state of genetic services:** • the implementation of a national registry of congenital anomalies will bring more visibility to the problem of congenital defects; • the National MoH is taking some actions to influence the provinces in developing comprehensive genetic services	**Current state of genetic services:** • brain drain of genetic health professionals due to unsatisfactory pay, lack of opportunities for career advancement • illegality of termination of pregnancy for foetal reasons impede reproductive options in case of genetic risks

Country	Strengths	Weaknesses	Opportunities	Threats
Argentina 		usually of an international agency • testing services in the private sector are more or less unregulated • while laboratories are certified by a state agency, participation in quality assessment programmes is voluntary and regulation very lax • no national guidelines and recommendations for the provision of medical genetic services including ethical guidelines		
	National policies to strengthen genetic services: • a plan for strengthening genetic services in the public sector which was adopted by the National MoH as a national policy for strengthening the network of genetic services in the public sector and for supporting training activities in medical genetics addressed to primary health professionals in disadvantaged areas of the country; recently, following the EU funded CAPABILITY project (2007-2009), special initiatives by national and provincial ministries of health have been started to improve genetic service delivery by	**National policies to strengthen genetic services:** • the National MoH does not have much leverage or resources to impose policies as each province has its own set of policies and budget	**National policies to strengthen genetic services:** • strengthening services via stimulating genetics research: in 2009, the Ministry of Science and Technology started a concerted effort with the National MoH and the private sector for research and development in genomic approaches in biotechnology and biomedical research, for which it has issued a number of calls for projects, with particular emphasis in public-private partnerships • building a national network of genetic services with proper regionalization and coordination • increasingly, health	**National policies to strengthen genetic services:** • fragmented, underfunded and understaffed public health sector services are unable to deliver the volume of needed services and will not be able to timely implement benefits derived from research in medical genetics and genomics • unchecked growth of for profit private health sector conspires against equitable care and prevention of genetic disorders

Country	Strengths	Weaknesses	Opportunities	Threats
Argentina 	increasing coordination and regionalization		professionals and patient organizations are becoming more assertive in claiming the right to health for patients with genetic disorders	
Brazil 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • "Live-born Declaration", a document issued by hospitals that in theory allows congenital anomalies present at birth to be registered systematically • participates in the ongoing ECLAMC • estimates on the prevalence of hereditary breast cancer and hereditary non-polyposis coli cancer available	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact on congenital and genetic disorders on health services • no established comprehensive population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders • although the "Live-born Declaration" document has been developed, due to ineffective implementation congenital anomalies are being underreported and the reliability of the data is questionable	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • the introduction of policies by the National MoH could help the growth of medical genetics in the country and organization of a functional network	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease burden • without effective primary interventions in place in the public health sector, the projected number of infants born with serious congenital and genetic disorders will increase over the next decades with important service implications • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-informed decisions on planned service development

Country	Strengths	Weaknesses	Opportunities	Threats
Brazil 	**Availability of key genetic services:** • mandatory newborn screening programmes available (covering PKU; CH in 27 states, SC in 14 states and CF in 3 states) • genetic counselling services established • provision of genetic counselling at primary care level • preconception care services available	**Availability of key genetic services:** • genetic testing services mostly available in urban areas at tertiary care level and in the private sector • the development of services in the private sector is opportunistic and mostly market-driven • genetic screening tests (except for newborn screening) are not available • genetic counselling services at primary care level only available in some regions and for specific disorders • no genetic services in the states of Amazonas, Amapá, Roraima, Rondônia and Tocantins • lack of MToP services especially in the public sector	**Availability of key genetic services:** • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level • providing testing services for others countries	**Availability of key genetic services:** • strong lobbies by religious groups against MToP • misconceptions regarding the need for medical genetics by physicians
	Access to genetic services: • services available in most large cities	**Access to genetic services:** • lack of universal coverage • inequitable access to services • genetic tests are often not available within the public health system due to insufficient number of services and scarce funding • restricted coverage of newborn screening (urban vs. rural areas) • disparities in the provision of	**Access to genetic services:** • for those who have private insurance it has become progressively easier to have genetic testing covered	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • excessive waiting lists in public health sector genetic services that implicitly lead to non-transparent

Country	Strengths	Weaknesses	Opportunities	Threats
Brazil 		services between urban and rural areas and between the south, eastern, north and western states		prioritization and rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps to recognize congenital and genetic disorders result in delayed referral
	Current state of genetic services: • recognition of medical genetics as a medical specialty • professionalization through the establishment of professional bodies and scientific societies' development of qualification standards (~200 physicians have been awarded with board certificates) • postgraduate programmes for laboratory services available • education programmes in medical genetics available for non-genetic health professionals • the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available, this includes the ability to undertake predictive testing for late-onset monogenic disorders	**Current state of genetic services:** • underfunded and understaffed public health sector services that are unable to deliver the volume of needed services are the norm • some Brazilian regions completely lack basic genetic service infrastructures to ensure care in genetics • there is no specific regulation for medical genetics services, that like all other medical services are regulated and supervised by National MoH and its specific agencies, particularly the ANVISA; some quality assessment programmes are available for laboratories, so that they comply with international standards; most private laboratories tend to undertake it voluntarily, not specifically for genetics, but for all testing offered.	**Current state of genetic services:** • possibility of future growth of the existing services with the new policies for genetics / rare diseases to be instituted by the National MoH	**Current state of genetic services:** • the genetics content in almost all medical schools does not cover even the needs of a general medical education.; therefore, most physicians do not recognize the genetic basis of diseases with which they are dealing and/or do not know how to refer to genetic services and/or do not give the deserved importance to the process of genetic counselling. • internal brain drain due to no job positions available for geneticists in the SUS which leads trained specialists to move to other practices / medical specialties where they can earn a living, and many never go into medical genetics again.

Country	Strengths	Weaknesses	Opportunities	Threats
Brazil 	• microarray and NGS introduced for diagnostic service purposes • pharmacogenetic testing introduced • national guidelines and recommendations for the provision of medical genetic services including ethical guidelines since 2003			
	National policies to strengthen genetic services: • the National MoH published a decree which proposes the creation of a *"Política Nacional de Atenção Integral em Genética Clinica no SUS"* (National Policy for Comprehensive Care in Clinical Genetics at SUS)	**National policies to strengthen genetic services:** • the decree is still waiting for implementation	**National policies to strengthen genetic services:** • strengthening services via setting research priorities: significant investments to fund research in medical genetics and genomics have been made • linking national institutes to further the development in human genetics and medicine via research	**National policies to strengthen genetic services:** • insufficient service structures: fragmented, underfunded and understaffed public health sector services that are unable to deliver the volume of needed services and will not be able to timely implement benefits derived from research in medical genetics and genomics
China 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • congenital malformations are monitored nationally via a hospital-based birth defect surveillance network and then reported to the National	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact of congenital and genetic disorders on health services • no established comprehensive	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • started to report data from hospital-based diagnostic testing laboratories to the HVP • biobanks established	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease burden • lack of national population-based epidemiological data

Country	Strengths	Weaknesses	Opportunities	Threats
China 	MoH annually. • marital & preconceptional health checks also involve screening for common genetic disorders, and data are updated to National Committee of Family Planning	population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders • systematic and accurate centralised national data collection is hampered by the marked absence of the correct identification at birth of children with congenital conditions • no data available on the prevalence of hereditary "late-onset disorders"		clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-based decisions on planned service development • without effective primary interventions in place in the public sector, the projected number of infants born with serious congenital and genetic disorders will increase over the next decades with important service implications
	Availability of key genetic services: • national newborn screening programmes cover more than 90% of the newborn population in the affluent eastern provinces • genetic counselling services established • provision of genetic counselling at primary care level • preconception care services available	**Availability of key genetic services:** • limited availability of genetic testing services • genetic testing services mostly available in urban areas at tertiary care level and in the private sector • genetic services cluster in the more affluent eastern and southern-eastern regions of China, poorer western and northern regions are underserved • newborn screening coverage falls well below 30% in the western provinces and is not available in Tibet • genetic counselling services at	**Availability of key genetic services:** • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level • provision of testing services for others countries	**Availability of key genetic services:** • the absence of professional regulation and guideline for molecular genetic services • insufficient number of specialists

Country	Strengths	Weaknesses	Opportunities	Threats
China 		primary care level only available in some regions		
	Access to genetic services: • the genetic service network has been fully established covers most of Primary cities (e.g. Beijing, Shanghai and Guangzhou) and Secondary cities (e.g. Chongqing, Chengdu, Hangzhou, Nanjing, Shenyang, Shenzhen, Tianjin, Wuhan and Xiamen) • national newborn screening programmes cover more than 90% of the newborn population in the affluent eastern provinces	**Access to genetic services:** • lack of universal coverage • inequitable access to services • most genetic tests have to be paid for out-of-pocket • restricted coverage of newborn screening (urban vs. rural areas)	**Access to genetic services:** • government has realized the limited access to basic genetic services • as the total birth defect rate is decreasing, government has the option to provide more political and financial support to improve the coverage of genetic services. • the stable national economic condition allows the expansion of the coverage for national medical insurance.	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • routine points of entry to genetic services at primary care level very limited • skill gaps in the recognition of congenital and genetic disorders result in delayed referral
	Current state of genetic services: • ~100 universities/colleges have medical schools. All these universities/colleges have genetics as compulsory, less than 10 universities, including Peking University, Tshinghua University, and Zhejiang University, have specially designed medical genetics instead of genetics; none of these set up separate courses for medical genetics • medical genetics or overall	**Current state of genetic services:** • medical genetics not recognized as a specialty, however, recognized as a sub-specialty in prenatal practice • the medical genetics professional in medical school has nothing to do with clinical genetic services. Genetic testing and genetic counselling are run independently in hospital, and generally are not combined and considered as	**Current state of genetic services:** • heavy governmental investment in next generation sequencing facilities (e.g. BGI), biobanking and global cooperation initiatives (BGI, HVP)	**Current state of genetic services:** • unmet needs: increasing numbers of people are demanding high quality genetic services • private sector development largely unregulated • underdeveloped clinical genetic services infrastructures will impede the translation process of technological advances generated in China into public genetic services

Country	Strengths	Weaknesses	Opportunities	Threats
China 	genetics is usually taught in the 2nd year of all medical undergraduate (including the courses for doctors, nurses, medical test technicians, public health personnel, medical administration personnel) as compulsory before the beginning of medical practice • the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available, this includes the ability to undertake predictive testing for late-onset monogenic disorders • microarray and NGS introduced for diagnostic service purposes • pharmacogenetic testing introduced • quality assessment schemes are available and the centres in hospitals are exposed to regular peer reviews; all clinical laboratories offering genetic diagnosis are required to meet the standards of Centre of Clinical Testing, National MoH; some private laboratories may also comply with ISO15189	"medical genetics" • in some small local genetic posts even genetic laboratory technicians may play the role of genetic counsellor, due to the lack of professional staff, although it is illegal • postgraduate programmes for biochemical, cytogenetic and molecular geneticists are not available • major hospitals provide only short-term professional training for non-genetic health professionals • brain drain/migration due to low salaries • majority of population remains underserved especially in the poorer western regions		

Country	Strengths	Weaknesses	Opportunities	Threats
China 	(Accreditation Criteria for the Quality and Competence of Medical Laboratories) and obtain accreditation from the CAP • national guidelines and recommendations for the provision of genetic services implemented since 2001 into the Maternal and Child Health Law • the National MoH has published a list of regulations concerning ethical issues			
	National policies to strengthen genetic services: • the Maternal and Child Health Law of the People's Republic of China includes genetic services recommendations for the provision of genetic services; in 2001, the China State Council published a State Council Order (No. 308) on implementing of the Maternal and Child Health Law; the law and the council order provide a detailed guideline for the development of maternal & child health care, especially in the area related to genetic diseases • National MoH has published a list of regulations concerning	**National policies to strengthen genetic services:** • the implementation of professional regulation & guidelines could be delayed and neglected in some under developed rural areas.	**National policies to strengthen genetic services:** • the increasing governmental and public awareness demands more legislative support for genetic testing.	**National policies to strengthen genetic services:** • the absence of national regulation for molecular genetic diagnosis greatly limits the implementation of tests .

Country	Strengths	Weaknesses	Opportunities	Threats
China	the management and implementation of human assisted reproductive technologies (e.g. PND)			
Egypt	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:**	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact of congenital and genetic disorders on health services • no established comprehensive population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders • systematic and accurate centralised national data collection is hampered by the marked absence of the correct identification at birth of children with congenital conditions • no data available on the prevalence of hereditary "late-onset disorders"	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • started to report data from hospital-based diagnostic testing laboratories to the HVP	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease burden • high prevalence of congenital and genetic disorders especially rare disorders • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-informed decisions on planned service development • underreporting birth prevalence of disorders • without effective primary interventions in place in the public health care sector, the projected number of infants born with serious congenital and genetic disorders will increase over the next

Country	Strengths	Weaknesses	Opportunities	Threats
Egypt 				decades with important service implications
	Availability of key genetic services: • national newborn screening programmes covering all 29 governorates (>90% coverage) • genetic counselling services established • preconception care services available	**Availability of key genetic services:**	**Availability of key genetic services:** • started to develop community genetic services based on counselling centres • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level	**Availability of key genetic services:**
	Access to genetic services:	**Access to genetic services:** • lack of universal coverage • inequitable access to services • coverage of genetic tests by the public sector is limited and services for poor people may be covered by donations from NGOs or individual charity • genetic services provided by the private sector have to be covered mainly by out-of-pocket payments • restricted coverage of newborn screening (urban vs. rural areas)	**Access to genetic services:**	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • excessive waiting lists in public health sector genetic services that implicitly lead to non-transparent prioritization and rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps in the recognition of congenital and genetic disorders result in delayed referral

Country	Strengths	Weaknesses	Opportunities	Threats
Egypt 	**Current state of genetic services:** • recognition of medical genetics as a medical specialty (the number of medical geneticists is estimated to be 100-150) • professionalization through the establishment of professional bodies and scientific societies' development of qualification standards • training courses on the detection of congenital and genetic disorders and referral to the community genetic clinics are available for nurses and physicians, in cooperation between the Ain Shams University department of paediatrics and the NRC; physicians working in the community genetic clinics receive condensed practical training courses of two months and can attend specialized courses. • the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available, this includes the ability to undertake predictive testing	**Current state of genetic services:** • medical genetics is taught only as part of the paediatrics curriculum • no postgraduate degree for any of the genetic laboratory specialties exists • inequity in the distribution of genetic specialists, positions in semi-rural, rural and remote areas remain unfilled. • underfunded and understaffed public health sector services that are unable to deliver the volume of services needed • no quality assessment schemes for genetic laboratories	**Current state of genetic services:**	**Current state of genetic services:**

Country	Strengths	Weaknesses	Opportunities	Threats
Egypt 	for late-onset monogenic disorders • pharmacogenetic testing introduced • genetic services to a certain extent integrated into the primary, secondary and tertiary health care, community genetic counselling clinics are a referral site between primary and tertiary care			
	National policies to strengthen genetic services: • policies and planning activities related to the provision of genetic services are included under the MoH&Ps five-year plan for the prevention and early intervention of disabilities • the MoH&P has established a national committee for community genetics leading to the development of 11 genetic counselling clinics in different Egyptian governorates	**National policies to strengthen genetic services:**	**National policies to strengthen genetic services:** • NRC has a Division of Human Genetics to strengthen genetic services	**National policies to strengthen genetic services:**

Country	Strengths	Weaknesses	Opportunities	Threats
India 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • data on the prevalence of genetic / congenital disorders is available from study of consecutive new births in a large number of secondary and tertiary care hospitals and a few hospitals in rural areas • data has also been gathered from genetic clinics in different centers, and an ICMR sponsored study on the prevalence of ß - thalassemia and other haemoglobinopathies from different regions of the country (Maharashtra, Gujarat, West Bengal, Assam, Karnataka and Punjab) • more recently the prevalence of hypothyroidism, CAH and G6PD deficiency is available from newborn screening studies	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact of congenital and genetic disorders on health services • no established comprehensive population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders, except the BDRI in Chennai (however BDRI data are mostly hospital-based data) • systematic and accurate centralised national data collection is hampered by the absence of national registry. • no data available on the prevalence of hereditary "late-onset disorders"	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • with the recent emphasis by the government of India on non-communicable diseases, opportunity exists for collecting data on congenital and genetic disorders; on February 6th 2013 the government launched the national *Rashtriya Bal Swasthya Karayakram* programme, which is a screening and early intervention initiative; under this programme children will be screened from birth to 18 years of age; at birth screening will be carried out for neural tube defects, Down syndrome, talipes, hip dysplasia, congenital cataracts, congenital heart disease, and retinopathy of prematurity; children aged 6 months to 6 years will be screened for nutritional deficiencies, skin disorders, hypothyroidism, sickle cell disease, and ß-thalassaemia; children from 6 to 18 years will be screened for developmental	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • while congenital and genetic disorders have become a major disease burden, the number of under-5 years deaths due to infectious diseases and malnutrition remain high at the national level, due to 80% of the population living in the rural areas. • underreporting of deaths due to congenital anomalies, poor universal clinical diagnostic services and inadequate surveillance and reporting systems • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely on the capacity to make evidence-informed decisions on planned service development • without effective primary interventions in place in the public health care sector, the projected number of infants

Country	Strengths	Weaknesses	Opportunities	Threats
			disabilities such as vision impairment, deafness, neuro-motor impairment, cognitive and language delay, and autism. In the first phase almost 270 million children will be covered.	born with serious congenital and genetic disorders will increase over the next decades with important service implications
India 	**Availability of key genetic services:** • newborn screening programmes established for five conditions, however, programmes not available in all states • PND is available in all care settings • genetic screening and carrier testing services available at primary, secondary and tertiary care level, however, availability is restricted • genetic counselling services established in major cities • preconception care services available	**Availability of key genetic services:** • genetic testing services concentrated in urban areas • genetic testing is costly and not covered by the insurance companies, although it is highly subsidized in the government institutions	**Availability of key genetic services:** • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level • providing testing services for other countries • increasing number of genetic units and genetic testing services primarily in the government sector. • as cost of genetic testing is low opportunity exists to extend these facilities to neighbouring countries.	**Availability of key genetic services:** • increasing establishment of genetic testing facilities in the private sector with high costs will make these less accessible to the low income population
	Access to genetic services: • in the urban areas genetic services are available in most cities • commercial laboratories have extended the reach of genetic testing services even to remote areas by establishing blood collection and dispatch centers.	**Access to genetic services:** • lack of universal coverage, social and private insurances usually deny coverage of genetic services on the grounds of pre-existing condition • inequitable access to services, the majority of patients and their families cannot afford out-of-pocket funding for testing services	**Access to genetic services:** • due to easy availability of trained scientific manpower genetic testing could be established in many government institutions • the excellent information technology services could be used creatively to provide genetic counselling services in remote areas through telemedicine and Skype	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • waiting lists in public health sector genetic services that implicitly lead to non-transparent prioritization and

Country	Strengths	Weaknesses	Opportunities	Threats
India 				rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps in recognizing congenital and genetic disorders result in delayed referral in the rural areas
	Current state of genetic services: • recognition of medical genetics as a medical specialty • postgraduate training programmes for biochemical, cytogenetic and molecular geneticists available • education programmes in medical genetics and genetic counselling are organized as courses and CMEs by various groups • NABL inspects and accredits the laboratories for genetic tests • ICMR has issued ethical guidelines for research in biomedical subjects in 2006 • ICMR has issued guidelines for the provision of medical genetic services • the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH,	**Current state of genetic services:** • not mandatory that every medical school should have a department of medical genetics for training purposes; there are only 5-6 medical schools that provide advanced training in clinical genetics, genetics is presently taught under various specialties like anatomy, physiology, pathology, paediatrics and internal medicine in the majority of medical schools • underfunded and understaffed public health sector services that are unable to deliver the volume of services needed • Indian population underserved, the number of genetic specialists insufficient (0.06 per million population) • 54 genetic units are presently running in different parts of India, this is considered	**Current state of genetic services:** • the demand for genetic centres in the private sector will drive genetic services in the country and provide jobs in the future	**Current state of genetic services:** • PhD students from institutions that are internationally recognized often migrate to the West following graduation • underdeveloped clinical genetic services infrastructures will impede the translation process of technological advances generated in India into public genetic services

Country	Strengths	Weaknesses	Opportunities	Threats
India 	and molecular diagnostic technology are available, this includes the ability to undertake predictive testing for late-onset monogenic disorders • microarray and NGS introduced for diagnostic service purposes • pharmacogenetic testing introduced	insufficient for a large country like India, some capitals of the 28 states do not have genetic services • India is a vast country and genetic counselling continues to be provided by general paediatricians, obstetricians, and physicians for the common genetic disorders occurring in Indian practice; there is a need to establish genetic centres with full laboratory support in the capital city of each state		
	National policies to strengthen genetic services: • substantial funds have been made available by the DST, DBT, CSIR and the ICMR to fund genetics/genomics research	**National policies to strengthen genetic services:** • there are no national policies/ guidelines in planning activities for provision of medical genetic services in India • genetics/genomics research is focused on the area of infectious diseases which afflict developing countries and less emphasis is given to congenital and genetic disorders • the government has established the Rural Health Mission to improve the health facilities in the rural parts of the country; the major focus has been on providing basic health needs and recently non-communicable diseases	**National policies to strengthen genetic services:** • NIBMG as an autonomous institution of the government of India established (2009) to create the necessary infrastructure to serve as an expert base for biomedical genetics/genomics	**National policies to strengthen genetic services:** • higher attention & priority given to infectious diseases, congenital and genetic disorders not a priority

Country	Strengths	Weaknesses	Opportunities	Threats
India		have been included in their scope of work; the focus is on diabetes, hypertension, coronary artery disease rather than congenital and genetic disorders		
Oman	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • strong hospital-based registries/surveys that provide figures on the birth prevalence of congenital anomalies • provides data on genetic disorders for CAGS	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no established comprehensive population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders • systematic and accurate centralised national data collection is hampered by the marked absence of the correct identification at birth of children with congenital conditions due to skill gaps • no data available on the prevalence of hereditary "late-onset disorders"	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • empiric national data on birth prevalence for congenital and genetic disorders to assist Oman in planning future medical genetic health services	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease burden, autosomal-recessive disorders are common • without effective primary interventions in place in the public health care sector, an increasing number of children born with congenital/genetic disorders will move into adolescence and adult life in the next years with important service implications
Oman	**Availability of key genetic services:** • mandatory national newborn screening programme available (covering 1 disorder)	**Availability of key genetic services:** • genetic testing services mostly available in urban areas at secondary and tertiary care level	**Availability of key genetic services:** • increasing number of genetic testing services available • started to develop community genetic services based on	**Availability of key genetic services:** • lack of trained Omani genetic specialists

Country	Strengths	Weaknesses	Opportunities	Threats
	• genetic counselling services established • provision of genetic counselling at primary care level • community genetic services available in rural areas providing genetic screening, carrier testing and genetic counselling services (primarily for haemoglobin disorders) • "*Central Notification of Birth Defects and Congenital Disorders detectable at Birth"* monitoring system • preconception care services available	• the scope of currently available testing services is limited	counselling centres • increasing number of conditions covered by the national newborn screening programme	
	Access to genetic services: • universal coverage provided by the state	**Access to genetic services:**	**Access to genetic services:**	**Access to genetic services:** • genetic services are concentrated in the main cities • routine points of entry to genetic services at primary care level very limited • skill gaps to recognize congenital and genetic disorders result in delayed referral
Oman	**Current state of genetic services:** • recognition of medical genetics as a medical specialty including qualification obtained abroad	**Current state of genetic services:**	**Current state of genetic services:** • new National Genetic Centre established	**Current state of genetic services:**

Country	Strengths	Weaknesses	Opportunities	Threats
	• the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available • microarray and NGS introduced for diagnostic service purposes • pharmacogenetic testing introduced			
	National policies to strengthen genetic services: • *"National Programme for the Control of Genetic Blood Disorders"* based on a community genetic model for controlling haemoglobin disorders by offering screening and counselling since 1999. • systematic planned national development of genetic services outlined in the MoH's 5-year-plans since 2005.	**National policies to strengthen genetic services:**	**National policies to strengthen genetic services:**	**National policies to strengthen genetic services:**
The Philippines 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • a compulsary national newborn screening	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact on congenital and	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • there is increasing awareness on genetic services among	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • genetic disorders have become a major disease

Country	Strengths	Weaknesses	Opportunities	Threats
The Philippines 	programme increases the availability of national data on birth prevalence of covered congenital/genetic disorders; PBDS Project is a national registry and surveillance project in the country that also aims to increase the national data on congenital/genetic disorders	genetic disorders on health services • no established comprehensive population based congenital disorder surveillance systems or registries • no data available on the prevalence of hereditary "late-onset disorders"	the health professionals and the general public (i.e. tri media campaign for newborn screening)	burden • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-informed decisions on planned service development • without effective interventions in place in the public health care sector, the projected number of infants born with serious congenital and genetic disorders will increase over the next decades with important service implications
	Availability of key genetic services: • Newborn Screening Reference Centre established • national newborn screening programmes, more than 3000 newborn screening facilities being available throughout the country • genetic counselling services established • preconception care services available	**Availability of key genetic services:** • genetic testing services mostly available in urban areas at tertiary care level and in the private sector • genetic screening tests (except for newborn screening) are not available • carrier testing not available • limited availability of reproductive genetic services in the public domain • genetic services only available in major areas such as Manila, Cebu and Davao	**Availability of key genetic services:** • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level • plans to expand newborn screening by 2013, coverage will extend from 5 disorders to more than 20	**Availability of key genetic services:** • the legal restriction of abortion hinders the development of PND, PGD and MToP services in the public domain

Country	Strengths	Weaknesses	Opportunities	Threats
The Philippines 	**Access to genetic services:** • there are 4 newborn screening centers all over the Philippines located in the 3 major islands of the country: 2 in Luzon, 1 in Visayas and 1 in Mindanao; there are also trained geneticists in these centers; another 4 newborn screening centers will be established in other parts of the country; the newborn screening programme will set up 17 regional follow up centers for patients with a confirmed diagnosis	**Access to genetic services:** • lack of universal coverage • inequitable access to services • genetic services and testing have to be paid for out-of-pocket • MToP not available	**Access to genetic services:** • genetic centres in tertiary care hospitals run telemedicine programmes for genetic consultations to overcome geographical barriers	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, out-of-pocket payments impact the ability of the poorer population to utilize services according to their needs • mostly the affluent urban upper-middle and upper classes can afford services • excessive waiting lists in public health sector genetic services that implicitly lead to non-transparent prioritization and rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps to recognize congenital and genetic disorders result in delayed referral
	Current state of genetic services: • the Philippine Paediatric Society has included genetics as a core topic in its curriculum for all medical schools • recognition of medical genetics as a medical specialty • the Department of Paediatrics, PGH offers a 2-year fellowship programme in	**Current state of genetic services:** • underfunded and understaffed public health sector services that are unable to deliver the volume of needed services • there are only seven trained medical geneticists for the whole country (0.06 per million population) • genetics is taught primarily in medical school as topics	**Current state of genetic services:** • a master's programme in genetic counselling is being offered since 2011-2012 at university level (at the UP-PGH); • increasing demand for genetic services • genetic research is a rapidly increasing field in the country	**Current state of genetic services:** • there is an urgent need for expansion and capacity building in medical genetics; the limitation in available medical geneticists not only severely hampers the ability to diagnose and manage hereditable disorders but also the ability to incorporate the benefits of

Country	Strengths	Weaknesses	Opportunities	Threats
The Philippines 	Clinical Genetics • postgraduate training programmes for molecular genetics are available • CME courses for non-genetic health professional provided by clinical geneticists • newborn screening integrated into the public health delivery system; all newborn screening laboratories are considered public health laboratories; guidelines and accreditation are run by the DoH • techniques available: conventional cytogenetic techniques constitutional, PCR, PT-PCR, metabolic biochemical testing • both internal and external quality assessment schemes, like the CEQA available; for the newborn screening, the Newborn Screening Laboratory-NIH-UP avails quality control samples bi-annually and proficiency testing samples quarterly from the CDC • all newborn screening centres undergo an initial accreditation and re-accreditation every 3 years	integrated in Biochemistry, Paediatrics, Internal Medicine and Obstetrics • formal postgraduate training programmes for biochemical genetics and cytogenetics are not available		genetic/genomics research into mainstream medicine • strongly affected by brain drain; the majority of trained original staff (of the IHG-NIH) have been absorbed by genetic laboratories overseas, and Philippine students going for PhD work overseas do not return

Country	Strengths	Weaknesses	Opportunities	Threats
The Philippines 	**National policies to strengthen genetic services:** • guidelines for the provision of newborn screening stated in the Newborn Screening Act of 2004 (RA 9288) • aside from the newborn screening law, other available national policies for the strengthening of genetic services in the country are focused on research and ethics: Operational Guidelines for Ethics Committees Reviewing Biomedical Research by UNDP/World Bank/WHO Special Programme for Research & Training in Tropical Diseases (TDR); Ethical Guidelines for Genetic Research with a Section on Stem Cell Research by the Philippine Council for Health Research and Development; Intellectual Property Code of the Philippines (Republic Act No. 8293).	**National policies to strengthen genetic services:** • infections remain to be the top priority of the DoH • improvement of genetic services are not on the priority list of policymakers	**National policies to strengthen genetic services:** • government investment in genomics • the Philippine Genome Center was put up under the UP Administration, and it includes a branch focused on health-related research	**National policies to strengthen genetic services:** • provision of basic genetic healthcare services to every region remains the biggest challenge in the country • lack of interest of policymakers in the field of genetics
South Africa 	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • limited hospital and community based epidemiology on	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • no data available on the impact on congenital and	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • very limited in the current situation in the country	**Congenital and genetic disorders burden: availability of national data/availability of epidemiological data on congenital and genetic disorders:** • while congenital and genetic disorders have become

Country	Strengths	Weaknesses	Opportunities	Threats
South Africa 	common congenital disorders is available. • provides data for neural tube defects (SABDSS) for the *International Clearing House for Birth Defects*	genetic disorders on health services • no established comprehensive population based congenital disorder surveillance systems or registries that document the birth prevalence of congenital and genetic disorders • stopping the SABDSS started in 1988 and involved the routine, systematic collection of data regarding congenital and genetic disorders from 15 hospitals in the country; however, such data collection ceased in 2005 and currently only data for neural tube defects is collected • systematic and accurate centralised national data collection is hampered by the marked absence of the correct identification at birth of children with congenital conditions • no data available on the prevalence of hereditary "late-onset disorders"		major disease burden, the number of under-5 years deaths due to infectious diseases, particularly HIV/AIDS and TB, remain high • underreporting of deaths due to congenital anomalies, due to poor universal clinical diagnostic services and inadequate surveillance and reporting systems • lack of national population-based epidemiological data clearly impairs health policy decision-makers' abilities to assess the impact of congenital and genetic disorders, which in turn impacts severely the capacity to make evidence-informed decisions on planned service development • without effective primary interventions in place in the public health care sector, the projected birth prevalence of infants born with serious congenital and genetic disorders will remain high over the next decades with important but unrecognized implications for mortality and morbidity and service needs.

Country	Strengths	Weaknesses	Opportunities	Threats
South Africa 	**Availability of key genetic services:** • medical genetic laboratory services are available in academic/ tertiary institutions and the private sector. • genetic testing is available for those accessing the public health sector by referral of specimens to the academic/tertiary institutions • limited genetic counselling services are established in academic institutions. • limited preconception care services available	**Availability of key genetic services:** • no mandatory national newborn screening programme	**Availability of key genetic services:** • increasing number of genetic units and genetic testing services primarily in the private sector or at tertiary care level • providing testing services for others countries	**Availability of key genetic services:** • the ambitious approach in the 1990s trying to offer medical genetic services including counselling services to the public through primary care was thwarted by the HIV/AIDS epidemic which forced the National DoH to shift priorities • the stagnation of the academic/tertiary clinical and laboratory services due to diminished political will, commitment and financial, structural and human resources
	Access to genetic services: • access to testing services is available through referral of specimens to academic/ tertiary laboratories.	**Access to genetic services:** • inequitable access to clinical services impacting the ability of the poorer population to access services	**Access to genetic services:** • limited in the current climate	**Access to genetic services:** • inequitable genetic services due to limited geographical accessibility, and limited affordability of services impacts the ability of the poorer population to utilize services according to their needs • excessive waiting lists in public health sector genetic services that implicitly lead to non-transparent prioritization and rationing of services • routine points of entry to genetic services at primary care level very limited • skill gaps to recognize congenital and genetic

Country	Strengths	Weaknesses	Opportunities	Threats
				disorders result in delayed referral •
South Africa 	**Current state of genetic services:** • recognition of medical genetics as a medical specialty • the basic testing techniques required to diagnose congenital and genetic disorders, chromosomal analysis, including FISH, and molecular diagnostic technology are available, this includes the ability to undertake predictive testing for late-onset monogenic disorders • pharmacogenetic testing introduced	**Current state of genetic services:** • underfunded and understaffed public health sector services that are unable to deliver the volume of needed services	**Current state of genetic services:** • limited in the current climate	**Current state of genetic services:** • implosion of genetic services in the public domain due to very limited political will, commitment and funding.
	National policies to strengthen genetic services: • South Africa has the constitutional, legal and regulatory framework in place to enable it to develop cogent medical genetic services to meet its health needs	**National policies to strengthen genetic services:** • the national lack of political will and commitment, and probably the ability to implement its own constitutional, legal and regulatory framework	**National policies to strengthen genetic services:** • the constitutional, legal and regulatory framework is more than adequate to meet the country's current medical genetic health needs	**National policies to strengthen genetic services:** • the continuing lack of commitment to the countries constitutional, legal and regulatory framework

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Abstract
Due to the epidemiological transition in the emerging economies of China, East Asia, India, Latin America, the Middle East and South Africa, these economies are dealing with an increasing proportion of morbidity and mortality due to congenital and genetic conditions, and a rising need for genetic services to improve patient outcomes and overall population health. For this reason, they are facing the challenge how to ensure the successful translation of genetic/genomics laboratory and academic research into quality assured pathways, and to develop a service delivery infrastructure that leads to equitable and affordable access to high quality genetic/genomic testing services. The GenTEE international project is intended to inform policy decisions for the challenges of delivering equitable high quality genetic services and to promote international collaboration for capacity building.
A standardized survey has been carried out, that is the first of its worldwide that allows comparison of services internationally across a number of key dimensions by using a core set of indicators, selected by the GenTEE consortium for their relevance and comparability.
To date, the GenTEE project has completed its survey that maps the current state of genetic services in the participating countries and identifies current drivers, barriers and opportunities for genetic services development. The results show that there is no equitable access to genetic services in all countries mainly due to financial barriers (underfunded fragmented public services, out-of-pocket expenses tend to be the norm for genetic testing services), geographical barriers (concentration of services in main cities) and skill gaps, resulting in inequitable services or delayed access. The development of services in the private sector is opportunistic and mostly technology and market driven. There is a marked lack of standard operating procedures and agreed quality assessment processes for new technologies. An international collaborative networks can provide support for capacity building and help to strengthen the provision of quality genetic/genomic services in emerging economies.

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