European Commission

JRC CONFERENCE AND WORKSHOP REPORT

Integrating genomics into personalised healthcare: a science-for-policy perspective

My genome: our future

JRC F.1, JRC F.7

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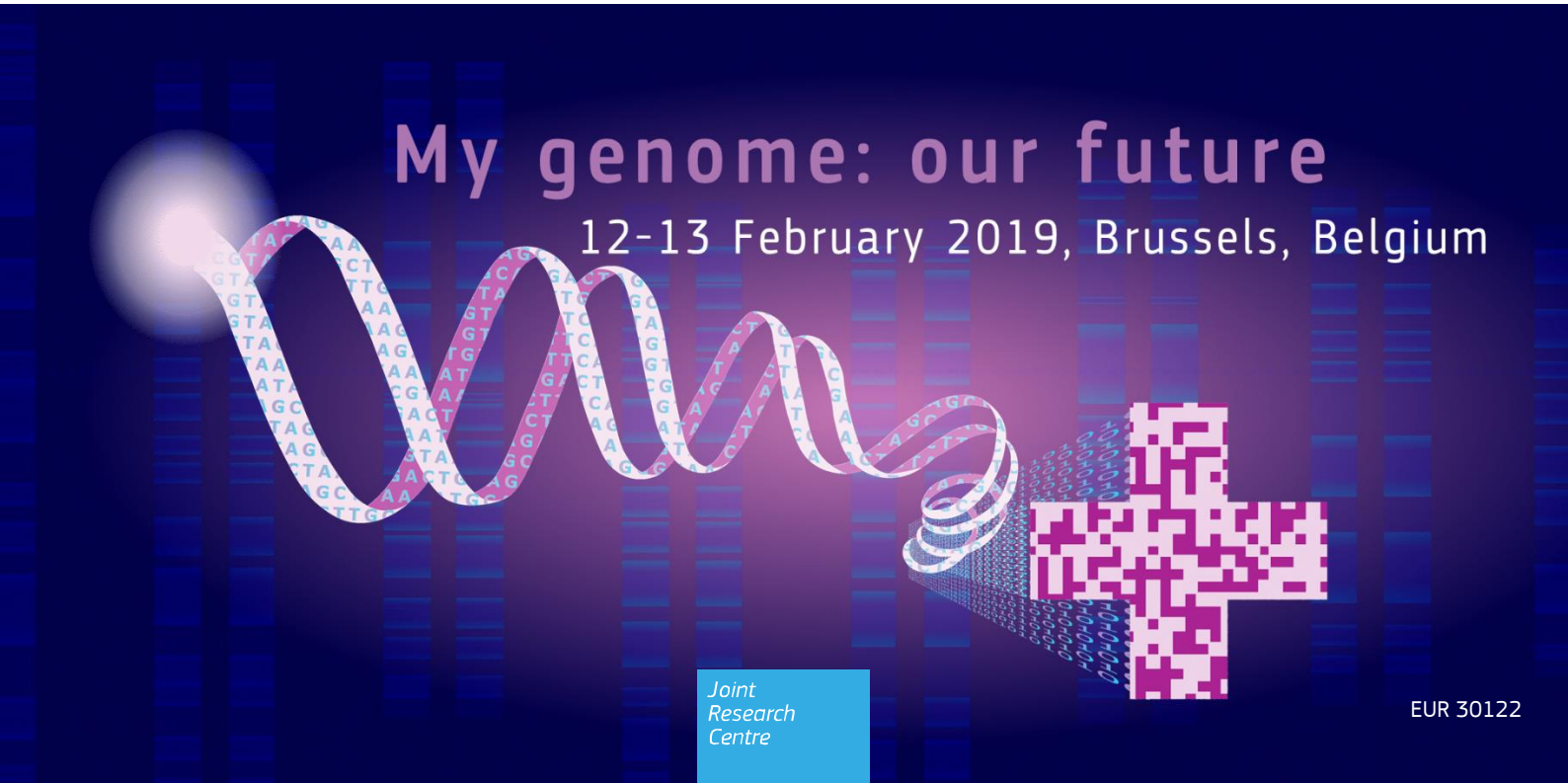

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Contents

Foreword

The latest developments in genomics have huge potential to impact the whole population and carry enormous ethical implications. These developments will also directly affect the sustainability of European Union (EU) healthcare systems. To discuss them, there is a need for more and better structured dialogue in the area of genomics between representatives of research and innovation community, healthcare professionals, public authorities and civil society.

With this in mind, the Joint Research Centre (JRC) took the initiative to organise a high level scientific conference, entitled 'Integrating genomics into personalised healthcare: a science-for-policy perspective', to promote cross-disciplinary interactions between specialists in genetics and genomics, health professionals, decision-makers, patient organisations and other relevant stakeholders. The conference aimed at bringing together experts from around the world to discuss the potential of genomics in the prevention, diagnostics, and therapy for cancer and the new opportunities it may bring for citizens and patients in the EU.

Many EU and national initiatives are blooming in this area. In 2018 the European Commission (EC) issued a Communication on 'Enabling the digital transformation of health and care in the Digital Single Market; empowering citizens and building a healthier society'. The three key priority pillars are:

1. citizens' secure access to and exchange of health data across borders, led by Commission's Directorate-General for Health and Food Safety (DG SANTE),
2. health data pooled to advance research, disease prevention and personalised medicine, led by Commission's Directorate General for Communications Networks, Content and Technology (DG CNECT), and
3. digital tools and data for citizen empowerment and person-centred healthcare, led by Commission's Directorate-General for Research and Innovation (DG RTD).

The majority of Member States (MS) has subscribed to a Declaration for delivering cross-border access to their genomic information and for bringing together infrastructure and expertise supporting a shared common goal to make one million genomes accessible in the EU by 2022. This declaration is in line with intention of the European Commission to adopt a Communication including, inter alia, 'supporting data infrastructure to advance research, diseases prevention and personalised health and care in key areas including rare, infectious and complex diseases'.

In addition, the Commission adopted on 6 February 2019 a Recommendation to establish a format for a European Electronic Health Record Exchange.

The JRC is already contributing to some of these initiatives; this conference and report are one example. It intends on continuing and is currently focusing on pillars 1 and 2 of the 2018 EC Communication.

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Abstract

The Joint Research Centre organised a high-level scientific conference, entitled 'Integrating genomics into personalised healthcare: a science-for-policy perspective', on 12-13 February 2019 in Brussels. This flagship event addressed, inter alia, current and future genome-based screening and diagnoses schemes in terms of quality assurance and implications for individuals, patients, the health care sector and society at large.

The focus of the event was on supporting patient-centric policies, covering important aspects such, data privacy and ownership and security of the genetic information produced, ethics and the impact of the genomics market on healthcare systems. The conference highlighted challenges to the implementation and the uptake of genomics-based screening and diagnosis in health systems and mapped the appropriate actions.

The event highlighted the strong disposition of the European Commission to foster integration of genomics into personalised healthcare and underlined the genomics potential to revolutionise healthcare in several ways. Moreover, it highlighted the European Commission ongoing actions to avoid that the 'potential' remains as such, by:

- increasing awareness amongst policy makers;
- supporting voluntary coordination mechanism of national, regional, and local public authorities to link the ongoing genomic medicine initiatives;
- supporting the development of technical specifications to secure access and cross-border exchange of genomic datasets and to facilitate responsible sharing of information;
- supporting a coordinated data governance framework;
- promoting the use of open standards and data management systems;
- facilitating the creation of the right environment for the uptake by reaching out and communicating to citizens, as part of the EU culture of trust and transparency.

The main messages highlighted during the conference, where special attention needs to be focused (see chapter 3), are:

- **A**ccess to data that have to be regulated, ensuring protection and responsible sharing;
- **B**etter economy tailored assessed and able to fund research;
- **C**apitalise on existing infrastructure, to take full advantage of the existing potential in terms of research and coordinated actions;
- **D**evelop education programmes not only for professional scientists but also to form well-informed citizens;
- **E**ffective regulation, that is the core for development in the field;
- **F**orging ties with society, engaging and empowering citizens.

1 Introduction

The rationale and the drivers behind organising this event stem from the plethora of, sometimes stand-alone, activities and an obvious need for knowledge management and seamless integration at European level. Within the JRC, for example, the following activities set the scene to trigger a more integrated approach:

- The European Commission Initiative on Breast Cancer (ECIBC) that will be made available to the Member States in 2020;
- It will also deliver a methodological blueprint for producing evidence-based guidelines and quality assurance criteria in healthcare for other cancer sites and chronic disease paradigms;
- Looking at the policy implications of scientific developments in 'omics' and in particular, 'genomics'. Here the focus is on the translation of genomics from research applications to health applications, covering prevention, diagnostics and therapeutics;
- A new JRC transversal project in liaison with DGs CNECT, SANTE and RTD. This supports the implementation of the Communication on 'enabling the digital transformation of health and care in the Digital Single Market; empowering citizens and building a healthier society' and on the Member States' Declaration for delivering cross-border access to their genomic information and for bringing together infrastructure and expertise supporting a shared common goal to make one million genomes accessible in the EU by 2022.

With other activities brought to the fore, the conference addressed the needs from both policy and societal perspectives and world-renowned experts were invited to address specific areas vital for integrating genomics into personalised healthcare.

A Scientific Steering Group was established to help elucidate the programme and to identify and to target key speakers and moderators.

The conference was held at the NH Bloom Hotel in Brussels on 12-13 February 2019. It brought together experts from around the world to discuss the potential of genomics in the health sector namely on prevention, diagnostics, and therapy for breast cancer but also in the field of rare diseases and other conditions.

The conference aimed at promoting cross-disciplinary interactions between specialists in genetics and genomics, health professionals, decision-makers, patient organisations and other relevant stakeholders. The focus was on supporting patient-centric policies, covering important aspects such as quality assurance, data privacy and ownership and security of the genetic information produced, ethics and the impact of the genomics market on healthcare systems.

The conference consisted of two days and was structured into four sessions, reflecting three main themes.

The three themes were:

1. Theme I: Genomics for health: the breast cancer example

What is 'genomics'? How does it apply to health? What's in it for me – as a citizen?

Starting from the use of genomics, in the field of breast cancer, the theme was meant to stimulate a broader discussion to illustrate the evolution of genetic testing and gene panels, with a particular focus on:

- how this facilitates bridging the application of whole-genome sequencing into clinical practice;
- how genomics can support better diagnostics, targeted treatments, disease monitoring and clinical follow-up.

2. Theme II. The public health perspective

What are the opportunities and challenges of implementing genomics approaches in public health? Who are the actors, and what are the actions that are needed for this implementation to succeed?

The theme was meant to foster a debate on public health topics like evidence-generating healthcare, harmonisation, reproducibility, data interoperability, quality and security in the context of genomics.

It also intended to map out how the process of translating genomic data into the clinical setting could be facilitated, and explain the impact that the genomics market has on healthcare systems.

3. Theme III. The citizen and patient perspective

How is genomics experienced by people, as patients, consumers, citizens? How will the citizen and the patient be approached and involved? How is the individual's information valued, and who will use and have access to it?

The theme intended to propose a consideration and reflection on regulatory and ethical challenges with a particular focus on the societal and legal implications for the citizen. It was meant to highlight the importance of literacy, both from citizens, patients and health professionals, and discuss ways to mobilise citizen engagement.

After the 'Institutional Addresses' and the scene setting, four scientific sessions were held over the 2 days. Each session consisted of one or two introductory speeches by top experts in the field and were followed by a panel discussion with experts representing different stakeholders related to that field.

The four sessions were:

1. Setting the stage
2. Genomics - Opportunities and challenges
3. Public Health perspective
4. Citizen and patient perspective.

The conference was concluded by a final keynote presentation. In addition, two special flash talk sessions took place: one in collaboration with the Health knowledge and innovation community of the European Institute of Innovation and Technology (EIT Health) by two young awarded health innovators, and one in collaboration with DG RTD on genomics for rare diseases.

The conference programme was defined in collaboration with the Scientific Steering Committee that also supported the identification of world-renowned experts as keynote speakers and panellists to address specific areas vital for integrating genomics into personalised healthcare.

The complete programme of the event is in Annex.

The 259 participants represented civil society, patient organisations, pharmaceutical companies, academics, national health authorities, research institutions, health care sector.

Taking advantage of operational intelligence, the conference used the services of One Tech to manage the participation, the social activities and the real time feedback of participants, as well as the social media interaction with the conference.

This document reports the outcome of the discussions in chapter 2, and has a final chapter dedicated to the recurrent key messages that have been identified during the conference. These messages are:

- **A**ccess to data that have to be regulated, ensuring protection and responsible sharing;
- **B**etter economy tailored assessed and able to fund research;

- **C**apitalise on existing infrastructure, to take full advantage of the existing potential in terms of research and coordinated actions;
- **D**evelop education programmes not only for professional scientists but also to form well-informed citizens;
- **E**ffective regulation, that is the core for development in the field;
- **F**orging ties with society, engaging and empowering citizens.

Reference to these messages is clearly highlighted in the conclusions of each session, where they are pertinent, and they are extensively discussed in chapter 3.

2 Conference contents

2.1 Institutional addresses

The conference was opened with two institutional addresses made by Mr. Vladimír Šucha, Director-General of the DG JRC and Mr. Martin Seychell, Deputy Director General of DG SANTE.

Both highlighted that there is a strong disposition of the European Commission to foster integration of genomics into personalised healthcare and both underlined that genomics has the potential to revolutionise healthcare in several ways. For example, Mr. Seychell said that genomics has the potential to revolutionise healthcare by leading to the development of more targeted and personalised medicines, therapies and interventions, by enabling better diagnostics and boost prevention. However, according to both, there are clearly challenges that are clearly of concerns (many of them were already mapped and identified in last year's Commission Communication 'Enabling the digital transformation of health and care'[1]). These challenges have been reported by Mr. Seychell in the form of 'lacks', as:

- lack of an appropriate technical infrastructure in the EU;
- lack of common agreed protocols to enable the data to be shared and understood by researchers and authorities in different settings;
- lack of clarity in the context of ethical and legal implications of genomics;
- lack of understanding from the general public, leading to scepticisms and fears in sharing of genomics data;
- lack of understanding on how to assess long-term implications of genomics (including pricing and reimbursement of personalised medicine) to ensure healthcare system sustainability.

According to Mr. Seychell, the combination of all the above 'lacks' might lead to a bigger one, namely the lack of genomics uptake by Member States, in their national policies, as well as in the healthcare settings, in general. In fact, the latest developments in genomics have huge potential to impact the whole population but they carry enormous implications, including in ethical and economic contexts. Until these issues are not addressed, the potential for genomics to accomplish more personalised, patient-centred healthcare in the EU will not be realised. Mr. Šucha and Mr. Seychell mentioned that 'the elephant in the room' is how genomics is expected to affect the sustainability of EU healthcare systems, especially through the use of Artificial Intelligence (AI), which is the other contemporary disruptive technology. According to Mr. Šucha, the application of AI to healthcare is crucial to make more efficient use of scare resources and genomics is at the heart. A proper framed health technology assessment could be a very useful tool in this respect: sharing expertise, resources, and workload in assessing such technologies is therefore seen as a must. Mr. Šucha used the terms 'interdisciplinary' and 'sharing' as keys to success.

Mr. Seychell emphasised the need to increase awareness among policy makers themselves, so that they engage with this topic and are encouraged to shape it for the better rather than resist or ignore it and to avoid that policy-making might become the bottleneck to the advancement and integration of genomics into personalised healthcare. The 1+ Million Genomes initiative, as part of the Declaration of cooperation towards access to at least 1 million sequenced genomes in the European Union by 2022[2] launched last year, led to great momentum and proved that there is a growing willingness in the EU to engage with and push ahead the area of genomics. Similar initiatives are ongoing outside the EU, like in China. Such willingness must be nurtured with the strong awareness of need of evidence that can help in the difficult task of remodelling the healthcare systems and properly guide the correlated investment calculated by policy makers. New and better quality data are needed, coupled with better use of existing data. For this, more targeted methodologies and frameworks have to be developed. The European Commission is already active in this sense by:

- funding several projects, like the International Consortium for Personalised Medicine (ICPerMed) which works on generating policy-oriented evidences;

[1] The Communication is available at https://ec.europa.eu/newsroom/dae/document.cfm?doc_id=51628
[2] The text of the initiative is available at https://ec.europa.eu/newsroom/dae/document.cfm?doc_id=50964

- leading the three key priority pillars of Commission Communication on enabling the digital transformation of health and care: (1) citizens' secure access to and exchange of health data across borders (led by DG SANTE); (2) health data pooled to advance research, disease prevention and personalised medicine (DG CNECT); (3) digital tools and data for citizen empowerment and person-centred healthcare (DG RTD). DG JRC's transversal projects will contribute to pillars (1) and (2).

Mr. Šucha and Mr. Seychell also stressed the need to create the right environment for the uptake by reaching out and communicating to citizens: positive outcomes and success stories related to genomics should be advertised and promoted so as to facilitate understanding amongst the population. In particular, Mr. Seychell mentioned the model of organ donation as a good example: the 'genomic data donation' could become a new practice in which citizens become involved, take ownership, and play a role. Providing feedback to 'donors' on how their data would eventually be used would be an important part of this. This need fits in the larger need of building a culture of trust and transparency: guaranteeing protection of personal data, security of stored data and ethical use of data, as well as defining clear data ownership rules, are the only ways to be successful in this action. It is also essential to ensure that the notion of discrimination on genetic/genomic grounds is categorically excluded: genomics has the potential to reveal facts about a person's genetic make-up which he/she is not responsible for and which they may even be unaware of. Only by clearly protecting a non-discrimination principle in the EU, it is possible to build the necessary culture of trust in this technology. In these respects, the EU has already shown that it can lead the way in setting strong guiding principles, which can be used to create and guide a 'EU model for integrating genomics into personalised healthcare', made of clear principles that could be widely applied across all Member States. Such a model would set very high standards on how genomics data is used, accessed, and stored, with clear ownership and data protection rules that protect citizens, ensure that data is used for ethically sound purposes only and clearly define public health objectives of the data use.

To capitalise on this, the appropriate infrastructures to support these actions need to be in place rapidly: Mr. Seychell mentioned existing tools like high performance computing and the European Open Science Cloud as potential baseline in order to see concrete progress soon. In addition, the European Reference Networks have the potential to make a useful contribution to advancing the work on genomics.

Throughout all this, it was recalled that innovation, such as in the field of genomics, needs to be seen as a tool and not as an objective in itself, as it is valuable and meaningful when it contributes to improving the effectiveness of the health systems, their accessibility, sustainability and resilience, thereby benefitting patients, and citizens as well. Both of them agreed that if Europe wants to improve people's health and wellbeing, it is fundamental to reflect on how the crucial organisational and human factors are addressed too, to avoid that genomics or any other innovative solution leads only to extra costs and/or to situations where it will be not affordable for most systems and citizens.

Mr. Šucha indicated that integrating genomics into personalised healthcare in the EU needs to be a collaborative task, involving specialists, researchers, health professionals, decision-makers, patient organisations and other relevant stakeholders. According to Mr. Seychell, this cooperation must be built upon existing initiatives in genomics and personalised medicine.

2.1.1 Conclusions

There is a strong disposition of the European Commission to foster integration of genomics into personalised healthcare and both underlined that genomics has the potential to revolutionise healthcare in several ways. The European Commission is active to avoid that the 'potential' remains as such, by:

- increasing awareness amongst policy makers themselves, to avoid that policy making becomes the bottleneck to the advancement and integration of genomics into personalised healthcare;
- supporting voluntary coordination mechanism of national, regional, and local public authorities to link the ongoing genomic medicine initiatives;
- supporting the development of technical specifications to secure access and cross-border exchange of genomic datasets and to facilitate responsible sharing of information (**A**ccess to data);
- supporting a coordinated data governance framework necessary to facilitate Europe-wide large-scale processing of health and related data in compliance with the applicable data protection legal framework, in order to support shared health policy goals (**E**ffective regulation);

- promoting the use of open standards and data management systems to ensure interoperability of genomic and other health data with a view to enhance research on personalised medicine (**A**ccess to data);
- facilitating the creation of the right environment for the uptake by reaching out and communicating to citizens, as part of the EU culture of trust and transparency (**F**orging ties with society).

2.2 SESSION I: Setting the stage

This session illustrated the evolution of genetic testing and how the application of sequencing technologies led to the discovery of genetic variants and subsequently to the eventual characterisation of disease variants. This ultimately may lead to applications in clinical practice and this session will highlight how genomics can support better diagnostics, targeted treatments, disease monitoring and clinical follow-up.

Chair: Joris Vermeesh, University of Leuven, Belgium.

Co-chair: Marco Marsella, European Commission, DG CNECT, Luxembourg.

Rapporteur: Jacques Simard, Université Laval, Canada.

Keynote presenters:

- Ewan Birney, European Molecular Biology Laboratory - European Bioinformatics Institute (EMBL-EBI), United Kingdom;
- Sir John Burn, Newcastle University, United Kingdom.

Panellists:

- Peter Devilee, Leiden University Medical Center, The Netherlands;
- Denis Horgan, European Alliance for Personalised Medicine, Belgium;
- Mark Bale, Genomics England, United Kingdom;
- Peter Kapitein, Inspire2live, The Netherlands.

2.2.1 Genome sequencing and diagnostics

Genomics achieved tremendous technology advances during the past years, leading to substantial decrease in the cost of sequencing a genome - which might reach $100 within the next 5 years, if we believe the manufacturer. This would put medical genomics costs at or below other «routine » medical diagnosis (e.g. MRI scans). Indeed, most of the costs will involve consent, DNA sample acquisition and analyses rather than the sequencing itself.

Although there are recognized caveats and challenges of whole genome sequencing, namely, error rates, inadequate depth of coverage, copy number variation, imprinted genes, splice variants, telomeres etc., this approach has huge potential to improve health care delivery. The utility of genomics in medicine is observed in almost all disciplines and for several applications, including screening, early detection, diagnosis and personalized medicine.

An important current field of application are rare diseases. For suspected rare diseases, there are several questions raised by patients or their families: What's wrong? (Diagnosis); What's the future? (Prognosis); What's to be done? (Therapy); Why did it happen? (Aetiology); Will it happen again? (Recurrence risk); Will it be as bad? (Clinical burden); Are there any tests? Under such circumstances, a diagnosis would end the 'diagnostic odyssey' for patients, which is painful, emotionally draining and costly for the healthcare service.

Currently, a large body of evidence supporting the clinical utility of genome sequencing in a clinical genetics setting is accumulating. It has been reported that the Like for Like Study in Australia led to a 5-fold increase in diagnoses at 1/3 the cost to previous standards of care. Currently a similar approach has been rolled out in many countries including Denmark, Finland, France and United Kingdom. In the United Kingdom, the 100,000

Genomes project was launched in December 2012. One of its first goal was to sequence 100,000 genomes from United Kingdom National Health System (NHS) patients and their families focusing on those with rare diseases or common cancers. At the time of the conference, 118,489 samples have been collected including 32,827 from patients diagnosed with a cancer and 85,662 individuals related with a rare disease; the analyses and results for 56,622 genomes have been sent to NHS Genomic Medicine Centres. It is noteworthy to mention that they obtained about 20-25% actionable findings for rare disease.

2.2.2 Breast Cancer

Breast cancer represents a major global public health problem. Currently, it is estimated that 1 out of 8 women will develop breast cancer during her lifetime. Breast cancer affects more than 360,000 women per year in the EU and causes more than 90,000 deaths. The detection rate of breast cancer in women has risen sharply, especially in the first half of the 1990s, mainly due to the introduction of breast cancer screening in women between 50 and 70 years old.

Strategies for breast cancer prevention include screening, risk reduction medication and prophylactic surgery. To keep cancer management and prevention up to date, it remains important to closely monitor the ever-changing breast cancer burden due to changing lifestyles, ageing, the improvements of early detection and the impacts of over-diagnosis.

The effectiveness of breast cancer prevention can be substantially enhanced through targeting prevention at those most likely to benefit. Risk prediction is thus important for clinical management. Risk prediction models are crucial to determine whether a woman is at low, moderate or high risk which then guides the clinical recommendations for prevention and screening according to risk categories, such as the age at which screening begins, the frequency of screening and the benefit from MRI or chemoprevention.

Improving the predictive power of risk models is an important step toward targeted screening and prevention. Currently, high-risk women are primarily identified on the basis of family history and mutation screening of the Breast cancer type 1 susceptibility protein (BRCA1) and Breast cancer type 2 susceptibility protein (BRCA2) genes. This approach has the limitation of missing women without family history but with a significant genetic predisposition. Indeed, many other risk-associated alleles in other genes have now been identified through the multigene panels used in clinical and research contexts. Testing of these genes with unproven association with breast cancer risk leads to results that are largely un-interpretable and users of the test may be seriously misled. Some of the challenges encountered are related to determining which of these genes are truly associated with the risk of breast cancer; determining robust risk estimates for these variants/genes; determining the subtype-specific risks associated with these genes and whether these genes confer risks to other cancers.

A few large-scale case-control studies have been performed recently to evaluate the association of a number of genes typically included in the current gene-panels, the largest one being the Horizon 2020 EU funded Breast Cancer Risk after Diagnostic Gene Testing (BRIDGES) project, which sequenced 35 genes in over 60,000 breast cancer cases and 60,000 matched controls. The major goal of BRIDGES is to improve targeting of breast cancer prevention, both in the context of family cancer clinics and the general population, by taking into account genetic data in addition to lifestyle risk factor to determine individual risk. Preliminary results of this case-control study have confirmed the association of the known major breast cancer susceptibility genes BRCA1, BRCA2, PALB2 (Partner and Localizer of BRCA2), ATM (Ataxia-Telangiesctasia mutated gene) and CHEK2 (Checkpoint kinase 2) and have further refined the risk estimates for these genes.

The logistics of genome sequencing approaches for cancer are complicated: sample acquisition and DNA extraction are difficult to standardise, and tumours tend to be heterogeneous. Still, there is a clear clinical need of deciphering the genomics signature of the disease, in particular since the high and moderate risk genes are known to account for only a small proportion of breast cancer cases in the general population. Multiple common breast cancer susceptibility variants have been identified through genome-wide association studies. These variants confer small risks individually, but their combined effects, when summarized as a polygenic risk score (PRS), can be substantial. Such genomic profiles can be used to stratify women according to their risk of developing breast cancer, as well as better inform decisions on cancer risk management. The predictive power of risk prediction models has major implications for personalised risk-based screening and for risk-reduction interventions.

Currently genetic counselling is aimed at detecting and excluding high risk genes. In the future, we should aim at assessing individual risk using genetic information for common and rare variants as well as lifestyle risk

factors. In this context, anyone can be tested and there would be no 'positive' or 'negative' results. Non-genetic risk factors should be incorporated into risk prediction and modifiable risk factors should be taken into consideration. Thus, it will be possible to identify a much larger group of women at high risk of breast cancer with much greater precision than is currently possible, allowing a more effective use of available resources to women who would benefit the most from screening and preventive strategies.

2.2.3 Coordinated infrastructures

The numerous ongoing national and international cohorts will generate a huge amount of genomics data in the near future, which can reach hundreds of millions of genomes by 2030.

Several international organizations, initiatives, consortia have been established during the past two decades to properly manage the data. This includes developing:

- the necessary tools for curating gene variants based on their validated functional significance;
- best practices for the different steps of the process;
- strategies to facilitate clinical implementation of genomic medicine and evidence generation to support its sustainability.

The Global Alliance for Genomics and Health (GA4GH; www.ga4gh.org/) is an international, non-profit alliance formed in 2013 aimed at accelerating the potential of research and medicine towards the advancement of human health. Bringing together 500+ leading organizations working in healthcare, research, patient advocacy, life science, and information technology, the GA4GH community is working together to create frameworks and standards to enable the responsible, voluntary, and secure sharing of genomic and health-related data. All of their work builds upon the Framework for Responsible Sharing of Genomic and Health-Related Data. GA4GH develops standards for genomics and healthcare, embracing a federated approach and setting community standards early.

Another example is the InSiGHT database hosted by the Leiden Open Variation Database (LOVD), which records DNA variants re-sequenced in the genes that contribute to gastrointestinal cancer, especially for patients with the Lynch Syndrome. More recently, the GA4GH BRCA Challenge project has brought together the existing international Evidence-based Network for the Interpretation of Germline Mutant Alleles (ENIGMA) consortium expert panel, along with expert clinicians, diagnosticians, researchers, and database provider. The goal is to advance the understanding of the genetic basis of breast cancer, ovarian cancer and other diseases by pooling data on BRCA1/2 genetic variants (21000 of which approximately 7,250 have expert classifications) and corresponding clinical data from around the world (www.BRCAexchange.org).

Genomics England has put in place an IT infrastructure to link the genomics and genetic diagnosis data to real world data of the patients including prescribing data, health records, digital pathological data, hospital episode statistics and mortality data. The project communicates the information regarding the patient's condition to all participants who agree to receive it. Moreover, there is an option to be informed about the carrier status for non-affected parents of children with rare diseases. Thus, ethics and transparency values are crucial to protect the interests of participants. In this regard, a broad consent for translational research has been obtained and an oversight of who should have access to participant data has been established.

Genomic England will undertake detailed development work on a new service to enable genomic volunteers to pay for a personalised report of their genetic makeup. As part of this, and with the permission of these volunteers, the genetic data will be made available to researchers and scientists working on tackling some of our country's greatest health challenges. They will work with NHS and patient groups to lead the development of the service.

In brief, it is now clear that data sharing is essential to enable reliable estimates of variant pathogenicity. A multitude of national and international organisations are seeking to address this challenge. It is considered that pseudonymisation can enable safe linkage of molecular and clinical data and legislation will be needed to help secure data. Genomic medicine will provide the essential information for targeting of effective expensive treatments will drive adoption. Finally, point of care technology will impact on clinical care.

2.2.4 Challenges and targeted opportunities for integrating innovation into Europe's Healthcare System

Although Europe has numerous strengths due to its public healthcare systems, strong expertise in genomics and needed infrastructure, there are some weaknesses and threats to address related to the IT depth in some healthcare systems, the fragmentation of skills, the AI and Big Data skills and human resources capacity and significant translational complexity to assure socio-economic benefits. It will be important to take benefit of the large quantity of genomics, epidemiological and medical information collected in numerous ongoing cohorts, such as the United Kingdom Biobank in which 100000 participants will be MRI imaged.

In order to better to determine 'What is the framework to ensure that health innovation gets into EU healthcare systems at a faster pace?' while taking into consideration, the policy framework, the regional development, the allocation of resources, and the SME role, it is first relevant to define and to share a common understanding of what is 'an innovation'. This is a translation of knowledge and insight into value. Although value to patients and society is a moving target, it appears that only 'learning health care systems' capture value.

Many reasons can explain why the value of a technology changes over time including:

- new technology with similar or improved outcomes;
- clinicians and patients who gather experience with the new technology, use it more efficiently or effectively;
- different indications with higher or lower incremental treatment outcomes compared to its original indication (e.g. cancer metastatic →adjuvant cancer indications);
- price changes of technology or relevant alternative technologies. The value of a technology can and must be actively influenced by payers and health care providers.

There is no doubt that tailored, local solutions to address barriers specific to each country are needed and this must be supported by EU-level debate and initiatives. A policy framework is needed considering multiple ongoing regulatory initiatives, changing science, increasing data volume, variety and velocity as well as a shift of the reimbursement landscape. In this regard, traditional approaches are limited by being frequently limited to single use case, which are not highly scalable and reinforce silos. Home-grown tools also struggle to keep pace with industry best practices associated with high operational and opportunity costs. Moreover, the established solutions are often not open platforms involving incomplete electronic data warehouse models and inflexible approaches. A good example of such coordination is the development a policy framework for health technology assessment (HTA). Since January 2018, 96 Europeans groups are working together on this issue.

One of the most important challenge is related to the data issue, especially given the multiple sources and types of data (e.g. electronic health records, health insurance data, registries, patient and physician surveys, hospital data, biobanks, genomes, digital phenotypes etc.). It is crucial to assure the interoperability of all layers keeping in mind not only the technical layer but also the legal framework, the business process as well as the semantics. This integration will require multidisciplinary teams bringing together key experts and strong project governance. A major barrier for such an integration is the inherent difficulty for a 'common understanding' which is crucial for the interoperability of health care information.

An ongoing 'Lighthouse initiative' will look into three sets of policy areas:

1. liberate the data but do no harm taking into account collaboration, data sharing, public/private partnerships, transparency, privacy, and ownership;
2. bring it now considering clinical adoption, data curation, veracity, security and common standards;
3. prepare the future promoting public education, workforce skills, information and communications technology infrastructures for life sciences, and bioinformatics analyses.

2.2.5 The Million European Genomes Alliance (MEGA) project

The Million European Genomes Alliance (MEGA) project has been established to provide access of at least 1 million sequenced genomes in the EU by 2022. This declaration of cooperation was originally signed by 19 member-state countries: Czech Republic, Cyprus, Estonia, Finland, Italy, Lithuania, Luxembourg, Malta, Portugal,

Slovenia, Spain, Sweden, UK, Bulgaria, Greece, Austria, Croatia, Netherlands and Lithuanian. More countries have joined the Declaration since.

European national strategies and initiative plans are highly consistent in their perception on benefits, principles and aims. The major aims of the MEGA project are to:

1. integrate genomic data into electronic health records and use this to support routine clinical decisions and an evidence-based and sustainable health system;
2. engage citizens by supporting their engagement in the use of genomic data for their own health and ability to contribute to research for societal benefit;
3. foster collaborative multi-disciplinary research in a hybrid research/care model to address the areas of most clinical need and ensure system adoption;
4. support a precision medicine industry encompassing development of genomics, health and technology enterprises in each country as well as encourage investment of leading global pharma, biotechnology and technology companies.

MEGA will achieve these objectives by taking the following actions:

1. leverage and maximally utilise existing resources;
2. develop a federated 'knowledgebase' of genomic and health information;
3. surround these knowledge bases with platforms for clinical discussions, training, research, innovation & enterprise;
4. establish a cross-border network of expertise and v) create a platform for participant engagement.

In order to create results that have impact on inter-regions, participants, industry, economy and society, the main focus of the strategy is the transfer of technology and application via the 'instruments' designed via Commission or Member States collaboration (e.g. Spaces/Lighthouse projects) funded through public private collaboration. The Operationalization will require the establishment of a coherent set of projects that complement each other without being dependent. Moreover, it is essential to define a framework for projects that ensure that 'results' (or even state of the art technology) is fit for the purpose of the next step in the innovation cycle and complies with the end user demand.

There are numerous expected benefits for stakeholders, namely: reducing cost of development, enabling innovation, patient stratification for benefit and risk, determining faster safety and efficacy in high risk groups, optimise indications and accelerated access to medicines, new outcomes measures and effectiveness data, improved EMA and HTA decision making and the ability to define the impact of such decisions, optimising use of medicines through ongoing monitoring.

2.2.6 Conclusions

As the technologies have reached a point where they are implementable in a cost-effective manner, the potential of Genomics to improve healthcare has become obvious, and the main question now is no longer 'whether' but 'what should be done to implement genomics?' The scientific community has demonstrated that it can be done, the clinicians agree that it is needed, and engaging with patients to find out their perspective will surely show that they will want it.

Some challenges remain, besides the natural resistance against change; some technical, some practical (for example, a large proportion of patients is not treated in academic hospitals). On the other hand, a lot of opportunities can be found in existing activities and established infrastructures. These are the subject of the following sessions.

To respond to the challenges, **A**ccess to data, **B**etter economy, **C**apitalisation on existing infrastructure,

Development of education programmes, **E**ffective regulation and **F**orging ties with society are all topics necessary to be tackled.

2.3 SESSION II: Genomics - opportunities and challenges

This session discussed concepts such as evidence-generating healthcare, harmonisation, reproducibility, as well as data interoperability, quality and security.

Chair: Giorgio Stanta, University of Trieste, Italy.

Co-chair: Irene Norstedt, European Commission, DG RTD, Belgium.

Rapporteur: Rolf Apweiler, EMBL-EBI, United Kingdom.

Keynote presenters:

- Jan Korbel, European Molecular Biology Laboratory (EMBL), Germany;
- Barend Mons, Leiden University Medical Center (LUMC), The Netherlands.

Panellists:

- Rodrigo Dienstmann, Vall d'Hebron Institute of Oncology (VHIO), Spain;
- Stefan Fröhling, National Center for Tumor Diseases (NCT), Germany;
- Serena Scollen, European Life-Science Infrastructure For Biological Information (ELIXIR), United Kingdom.

Through the large-scale sequencing projects of cancer exomes/genomes, the most common genetic drivers of cancer (>5% frequency per tumour type) have been defined. This already allows the use of this information for personalised medicine, selecting the type of treatment based on the intracellular pathway affected by the genetic mutations, rather than the type of cancer.

2.3.1 Cancer molecular characterisation

The NCT MASTER program started in 2013, and registered exome and RNA sequencing from young adults with advance-stage cancers and patients with rare cancers to design molecularly stratified clinical trials. Since March 2016, this program became the joint German Cancer Consortium (DKTK) activity, whose aim is to stratify cancer patients into treatment baskets (based on molecular profiling and the identified affected biological pathways) within 28 days of enrolment. The molecular characterisation led to re-evaluation of the clinical diagnosis in about 5% of the cases, stressing the importance of the close interaction with pathology.

Comprehensive molecular profiling, using stratification approaches, are now needed to capture the rare driver mutations (85% of all hotspot mutations affect less than 5% of any cancer type in which they are found), as well as the complex, multifactorial biomarkers.

There is a significant potential for increased cohort sizes to study cancer. The prerequisites for this are (1) data sharing (2) standardized analysis protocols. There are various Europe-led projects relevant for this approach, including the GA4GH, the Million European Genomes Alliance, the Pan-Cancer Analysis of Whole Genomes (PCAWG) and the International Cancer Genome Consortium (ICGC). For large datasets being effectively shared during collaboration, it is important to consider whether data processing & sharing is compliant with the implementations of the General Data Protection Regulation (GDPR), and whether the data analysis tools can adequately deal with the different clouds.

An example of data sharing for improved understanding of the genomic and molecular basis of diseases is the GENIE program, an international pan cancer registry to improve clinical decision-making. The registry operates through regular uploads of data by partner institutions, which are then mapped to a common ontology and harmonised, while removing unnecessary personal health information. Clinical query and data access requests are sent by partners through a common portal leading to the publication of more than 20 publications using the Genomics Evidence Neoplasia Information Exchange (GENIE) data as control/reference cohort since 2017.

GENIE currently focuses on Genomics and clinical data (tumour type, histology, etc.), with the aim to add also other omics and phenomics data and share information as widely as possible to facilitate and inform interventional clinical trials, especially basket trials.

2.3.2 Genomic data sharing

The sharing of genetic data has value for research but also for patients, as long as they are properly informed, upfront, about the existing risks. Specifically, for cancer, Genomic medicine can be used to stratify patients (identifying those that require treatment), identify patients at risk that can benefit from monitoring, identify the drugs adequate for treatment and detect the subset of patients that may benefit from immunotherapy. For example, a recent EMBL project, involving the sharing of 1022 medulloblastoma genomes and exomes (mainly from Germany, Canada, United States), resulted in new clinical guidelines for paediatric brain tumour medulloblastoma.

For rare diseases, sharing of genomic data also helps generating fundamental insights on the genetic bases of the diseases, allowing to provide accurate advice to family members and improve the efficiency of the diagnostics for the patients.

In order to realise the full potential of genomics for patients in Europe, genomic profiling to enable discovery and clinical decision making needs to be more widely accessible. For this, interaction between clinicians and the research community is key. The genomic data need to be efficiently shared international, for which the appropriate incentives and global standardization are key.

A popular proposal for the international sharing of human genome data is a 'federation', i.e. the datasets from human research participants are stored locally, and made available through a system of linked national data clouds.

Large, complex and privacy sensitive datasets will increasingly 'stay where they are'. Consequently, analytics move to the data as a rule and data to analytics as the exception. In case data are about a particular citizen, that person is in principle in charge of the re-use of this data for research and other purposes. Clinically collected and self-reported health data are not technically different from research-created data and should be dovetailed for optimal use. This means that they should comply with the FAIR (Findable, Accessible, Interoperable, Reusable) principles – they should be findable, accessible, interoperable and reusable. There are challenges for this, in addition to the legal, organisational and technical hurdles: the process can be burdensome for the health care professional, who may see limited benefits from making the data available. There is also the technical debt of legacy data to consider, as well as the lack of a basic, broad purpose common infrastructure.

In order to achieve this, health data should be represented as 'digital objects', ranging in size from sequence to a particular concept (gene, variant, etc.) to modularly increased complexes. Digital Objects, with their own ID, metadata, operations info (including license etc.) are routed to each other via a Digital Object Interface Protocol (DOIP). Both data sets next to compute and the workflows dealing with them are again digital objects and then need machine actionable metadata. Knowledge discoveries, in this model, are driven by 'personal health trains', algorithms that 'drive' to where the data is made available (through the FAIR principles) and perform their analyses locally. In this metaphor, the 'train tracks' are secure environments, owned by public entities that grant and manage the access to the system. Certain existing databases can be seen as 'core resources' due to their central importance and should be globally endorsed and supported sustainably.

2.3.3 Genomic data infrastructures

Large, coordinated infrastructures are thus necessary for transition of human genomics data into healthcare. ELIXIR connects national bioinformatics centres and EMBL-EBI into a sustainable European infrastructure for biological research data. In the field of rare diseases, ELIXIR established a Registry of Rare Disease data resources and tools for data discovery and sharing, interoperability and training; the aim being to coordinate efforts to enable all people living with a rare disease to receive a diagnosis within one year. ELIXIR and GA4GH have a strategic partnership, working towards the development and implementation of standards, APIs (Application Programming Interfaces) and toolkits to be used throughout ELIXIR Nodes for human data discovery and access.

Currently, technical knowledge about how to manage data is in the research domain. Healthcare needs to tap into this through national initiative projects that would buy in that genomics will benefit healthcare. For this, the challenges faced include data ownership, the selection of driver projects in certain disease areas, training enough people able to handle data in a way to get most out of the data, the importance of patients seeing the benefits of data sharing, questions of quality and relevance of data for clinical utility, and finally the importance of a long-term sustainable database landscape as a foundation for life science progress.

2.3.4 Conclusions

In order to fully exploit the evidence-generating potential of genomics for healthcare, there is the need:

- to establish a coordinated, secure, federated environment (**A**ccess to data);
- to fully take advantage of the lessons learned and solutions developed from existing infrastructures (**C**apitalise on existing infrastructure);
- for the EU to take a lead on policy-framing and technical standards-setting on a global stage in collaboration with organisations such as GA4GH to enable responsible genomic data sharing (**E**ffective regulation);
- to draft minimum recommendations for EU-wide infrastructure to access and analyse genomic data standards, APIs, computational resources, tools and services, regulatory frameworks and training and capacity building programmes (**D**evelop education programs);
- for the development and implementation of standards that are fit for purpose and stable;
- for a long-term, sustainable, database landscape as a foundation for life science progress (**A**ccess to data);
- to implement this while gaining and keeping the trust of patients (**F**orging ties with society).

2.4 SESSION III: The public health perspective

The session mapped how the process of translation of genomics data to the clinical setting could be facilitated, and explain the impact the genomics market has on healthcare systems.

Chair: Tit Albreht, National Institute of Public Health (NIJZ), Slovenia.

Co-chair: Elke Anklam, European Commission, DG JRC, Belgium.

Rapporteur: Marc Van den Bulcke, Sciensano, Belgium.

Keynote presenter: Thierry Philip, Institut Curie and Organisation of European Cancer Institutes (OECI), France.

Panellists:

- Nazneen Rahman, Independent healthcare consultant, United Kingdom;
- Jacek Gronwald, Pomeranian Medical University (PMU), Poland;
- Jan-Ingvar Jönsson, Swedish Research Council, Sweden;
- Laura van't Veer, University of California San Francisco (UCSF), Helen Diller Family Comprehensive Cancer Center, United States.

2.4.1 The Cancer Mission

The history of European funding for cancer basic and translational research showed a tendency to be too dispersed. Efforts were made in Horizon 2020 should include more open source, foster global science and industry experiences, and be more open to innovation. Recently, a new tool has been proposed: the Mission, whose ambition is to be bold, activating innovation across sectors, across actors and across disciplines. In this frame, the Vision for 2030 is the long term survival of 3 out of 4 cancer patients in countries with well-developed healthcare systems. This will be achieved through a network of networks, covering aspects such as prevention, early diagnosis and screening, basic research, clinical research and outcome research.

To be successful the European Cancer Mission should ensure that its scope is overarched in an EC strategic plan, and that the Governance of the Mission has a clear definition by a mission board that includes

patient/citizen input, and is co-created/co-designed with the involvement of all stakeholders (a 'cancer parliament').

In the first OECI European Mission Working Party Meeting, it was proposed to foresee small numbers of Missions with specific goals. The Mission for paediatric cancers aims for a survival of 90% of affected children, with lowered toxic side-effects. For adults, the Mission aims towards different targets: lower cancer incidence, higher quality of life and survival and increased quality of rehabilitation of survivors. Primary prevention, in close relationship with National cancer plans, is thus a crucial component of the Cancer Mission.

The integration of Genomics in the missions is thus crucial, as is can impact on all important aspects: Prevention (personalized preventive measures), early diagnosis (e.g. blood circulating malignant cells), fundamental and translational research, and personalised medicine. Genomics can also contribute to the fundamental/translational research needed to better understand immune responses.

2.4.2 Prevention and identification of high-risk individuals

Although Cancer Predisposition Gene (CPG) Testing has been available for 25 years, these tests are still not successfully delivered to the most eligible individuals, leading to lost opportunities for better cancer management and prevention. One of the reasons for this failure is the complexity of establishing eligibility for the test. Many eligible patients are not referred to genetic counselling, or incur long delays. For example, BRCA testing eligibility is complex and variable across Europe. When efficient criteria are implemented, mutation detection increases, leading to cancer prevention which is practical and highly cost-effective.

2.4.3 Diagnostics and Personalised Medicine

The aim of personalized medicine is to achieve the most optimal outcome at the individual level, both for patients and citizens, by guiding the decisions taken by health care providers. Precision oncology uses available information to personalise the intervention and allows to either

1. escalate the treatment to increase efficacy or
2. de-escalate the treatment to reduce morbidity.

Genomics assays for diagnostics of early stage breast cancer are needed to add greater precision (compared to clinical and pathologic factors) and guide early therapy decisions. An important actor in personalised medicine is ICPerMed. ICPerMed is a platform connecting 42 European and international institutions to foster and coordinate research on personalised medicine. ICPerMed strives to establish its members as global leaders in personalised medicine research, and identified two important aspects for this:

- the stimulation of coordinated research to expand the medicine science base;
- the investigation of the benefits of personalised medicine for citizens and healthcare systems.

The 2018 ICPerMed conference highlighted existing best practice examples in personalised medicine research and implementation, including implementation for lung cancer care in clinical routine (the Network of Genomic Medicine in Germany), rare diseases and cancer (Genomics Medicine Sweden) and familial hypercholesterolemia.

In another example, clinical studies in Poland demonstrated the importance of genetic testing in the optimisation of prophylactics and treatment of patients with breast - ovarian cancer syndrome.

Diagnostics are the keys to precision medicine. However, there is zero consensus on the level of evidence a diagnostic must show to be accepted as clinically useful. This also makes it difficult to design the right coverage and reimbursement systems for these procedures. An important aspect in the implementation of personalised medicine is the need to demonstrate not only evidence but also cost-effectiveness of the approaches.

An example of a prognostic and predictive diagnostic test for early stage breast cancer patients that was brought from Science to Healthcare is the MammaPrint assay. MammaPrint is a microarray based assay that measures the expression of a panel of 70 genes. It is used to aid treatment decision by identifying for which patients the benefits of chemotherapy would outweigh the harm, following a randomized prospective clinical trial validating its clinical utility. MammaPrint is now included as standard of care in some cancer management guidelines, and some EU countries defined a financing (partial or full reimbursement) of the test.

2.4.4 Conclusions

In summary, there is a need to define a clear vision for the next decade in the fight against cancer. The basis for this vision should be prevention and early detection, linked to Public health genomics. Achieving a consensus on the implementation, as well as standards for evaluation, of genomics into healthcare requires a coordinated effort from the partners involved, including HTA organization, health care professionals, patients, patient advocacy groups and insurance companies. The long-term vision needs to include innovative medicines, and to link pathology with Artificial Intelligence driven genomics. This vision should be developed as a co-creation process EC and Member States.

Access to data, a **B**etter economy, **C**apitalising on existing infrastructure, **E**ffective regulation and Forging ties with society are fundamental for the achievements of the goals.

2.5 SESSION IV: The citizen and patient perspective

How is genomics experienced by people, as patients, consumers, citizens? How will the citizen and the patient be approached and involved? How is the individual's information valued, and who will use and have access to it?

This session discussed regulatory and ethical challenges and considerations, with a particular focus on the societal and legal implications for the citizen. It will highlight the importance of literacy, both from citizens, patients and health professionals, and highlight the ways to mobilise citizen engagement.

Chair: Peter Goodhand, Global Alliance for Genomics and Health, Canada.

Co-chair: Ioana-Maria Gligor, European Commission, DG SANTE, Belgium.

Rapporteur: Rita Schmutzler, Uniklinik Köln, Germany.

Keynote presenters:

- Bartha Knoppers, McGill University, Canada;
- Francesco Florindi, Biobanking and Biomolecular Resources Research Infrastructure - European Research Infrastructure.

Panellists:

- Effy Vayena, ETH Zürich, Switzerland;
- Bettina Borisch, University of Geneva, Switzerland;
- Jean-Pierre Hubaux, Ecole Polytechnique Fédérale de Lausanne (EPFL), Switzerland;
- Andres Metspalu, University of Tartu, Institute of Genomics, Estonia.

The efforts to properly share human genome sequences should be framed within the Universal Declaration of Human Rights, in particular Article 27: 'Everyone has the right freely to participate in the cultural life of the community, to enjoy the arts and to share in scientific advancement and its benefits'.

This declaration is legally actionable, and MS should use it to establish mechanisms for collaborative, open science. The contribution of ordinary citizens to scientific advancement should be stated as a human right, reorienting the debate from 'policing science' to 'governing society'.

2.5.1 Citizen engagement

When engaging with the citizens about personalised medicine, it is important to seek as much input from them as possible, including focus groups and surveys. It is also crucial to properly explain complex concepts in

personalised medicine, such as the different 'risk' levels and what is implied by 'intermediate risk'. The citizen is both the main target of biotech companies and other health-related business and needed for academia and research; however they are often the group with the least amount of influencing powers on all other stakeholders. It is important to represent the 'healthy citizen' in public health discussions - although there is a political will for inclusion, the citizen currently feels excluded.

An important concept in this context is Equity. This covers:

- ensuring a correct diversity when performing genomics research (e.g. current Genome-Wide Association Study (GWAS) discoveries are overwhelming performed using white European cohorts);
- distributive justice, to ensure that individuals and groups fairly benefit from the health benefits of personalised medicine;
- finding the proper balance between Individual Benefits (privacy) and Collective Benefits (sharing);
- the reduction of social inequities (solicitation, informed consent and return of results vs levels of literacy) and regional disparities (rural vs urban areas). Digital health and electronic forms of communications can potentially help reduce these inequities;
- an efficient reciprocity between health systems and the citizens (contribution of information as a patient to improve databases, resulting in better healthcare and screening).

2.5.2 Biobanking

Biobanking plays a crucial role in all aspects of human genome sequencing and personalised medicine, including all phases of medicine development and diagnostics. Biobanking is built on trust from the citizens, and can only work if that trust is properly managed and respected. Public funding of biobanks may be a way to build this trust.

Large biobanks projects (e.g. the Biobanks and BioMolecular Resources Infrastructure (BBMRI) or in Estonia) thus need to leave an important voice in their advisory boards to patients groups, to properly support the challenges of ethical, legal and societal issues linked to biobanking and the use of these samples for research. A biobank should be prospective, longitudinal and volunteer-based, and need clear access and consent rules to be open for scientific research. Information can be returned to the donor who agree to receive it, in the form of a 'polygenetic risk score', which requires proper and professional counselling to be interpreted correctly.

Some important current issues in biobanking include:

- the efficient utilization of existing biosamples (currently, only about 10% are being used for research);
- the development of a code of conduct to develop biobanking policies compliant with the GDPR. This also includes a need of exchange between patients and policy makers to eventually adapt the GDPR in order to include the particularities linked to genomics data (which is not the main subject of the current regulation);
- validating the in depth understanding of the consent form by donors (how 'informed' it really is);
- the incidental finding policies, which in the EU have been said to be too conservative;
- meeting the citizen expectations on the return of research results, which are constantly evolving;
- avoiding duplication and fragmentation across different biobanking initiatives.

Another important interface between the citizens and genomics is the expanding private sector of recreational genetics. A lot of citizens are customers, producing a wealth of information now accessed by biotech companies without a clear mechanism of sharing the profits back to society. The regulatory frameworks are not adapted to this 'social network'-type of research, leading to a form of surveillance capitalism where populations are harvested for their (genomics) data. It is important to build a solid legal framework based on the values and trust, communicating with the citizen not only for input but also to explain the work being done.

2.5.3 Protection of personal information

It is important to keep in mind that, even within a well-designed legal framework regulating the proper use of genomics information built on dialog with and trust of the citizens, genomics data is, like all personal and health-related data, a target of interest of organised crime. Security, and the protection against breaches and unauthorised use, must be an important component of the framework.

Experience has shown that de-identification of genomics data, to facilitate sharing for research uses, is impossible - genomics data is too rich in dimensions. It is important to rethink how data is protected in this context, and technologies are being developed such as:

- traditional encryption (that can protect the data when stored or in transit, but not when being used in computations);
- homomorphic encryption (that allows computation on encrypted data);
- trust vendors for trusted execution environments;
- blockchain and related technologies, that don't protect privacy but allow strong accountability and traceability, for example when logging operations in a hospital system.

2.5.4 Conclusions

When properly fostered, the implementation of personalised medicine is expected to improve many aspects of public health: precision diagnostics (e.g. for rare diseases), precision treatment (e.g. cancer therapies), personalised prevention (e.g. against common diseases) and pharmacogenomics (i.e. anticipate drug responses). Efficiently integrating genomics into a public healthcare system produce a virtuous cycle of 'evidence-generating medicine' where new samples improve the existing knowledge. Evidence-generating medicine comes with its own challenges, which include:

- proper awareness of both doctors and patients (**D**eveloping education programs);
- correctly empowering the citizens to manage their own health (**F**orging ties with society);
- the need of better databases that allow the discovery of new associations between DNA variants and diseases (**A**ccess to data);
- the resources needed to constantly update and implement the evidence thus generated (**B**etter economy).

3 Main conference messages

This section summarises facts, opinions, discussions points and ideas put forward during the meeting. We credit the speakers, panel discussants and participants in general for the points detailed below

3.1 Access to data

In the context of these discussions 'Data' is mentioned often as:

- data sharing, e.g. making genomic data and other health related data such as health outcomes useable for research;
- data protection, i.e. legal control over access to and use of genomic (or other types of health) data;
- data content and metadata, i.e. 'data about data', in this case a set of descriptive data that gives information about the genomic data.

The debates left no doubt that sharing of genomic data is essential to reap its promise and potential. Many national and international organisations, such as Global Alliance for Genomics and Health (GA4GH), are focusing on providing a 'responsible' sharing framework for genomic and health-related data based on a federated approach and a set of common standards.

Interoperability of these sets of data comes with challenges, as 'genomic data' (in the form of genomes or panels of genotypic variations) evidently consists of multiple types of data and associated meta-data. These data can be linked to electronic health records, health insurance data, registries, patient and physician surveys, hospital data, bio-banks, digital phenotypes, etc. The interoperability of all these layers of objects was seen as crucial, during the conference discussions; the interoperability of data and infrastructure technicalities needs to be addressed but also the legal frameworks, the business processes and the semantics. Multidisciplinary teams of key experts, supported by strong governance projects will be needed for this. This broad and inclusive approach was discussed as the most efficient solution to achieve the 'common understanding' needed for a viable integration of genomics data and practices into healthcare.

There are four 'sets' of policy areas (three of them indicated by the Electronic Components and Systems for European Leadership (ECSEL JU) Lighthouse initiatives, see section on 'Infrastructure') needed for access to data to materialise:

1. A policy area that democratises responsible sharing of genomic data, that liberates the data but does no harm, underpinned by values such as responsible collaboration, responsible data sharing, responsible public/private partnerships, transparency, privacy, and ownership. There are numerous cohorts projects being set up; it has been estimated that there can be hundreds of millions of genomes by 2030. We will soon suffer the lack of agreed policy governance.
2. A policy area that supports and protects the use of the data above in clinical settings to the benefit of individuals and patients. Issues such as data curation, veracity, security and common standards will need to be addressed. For example, the United Kingdom Biobank will image 100,000 participants with MRI and again federation of data appeared as a widely accepted model to avoid privacy issues and national boundaries. Analytics move to the data and not the other way around. However, a series of agreed rules need to be set, like rules to define the Creation of Digital Objects and Digital Object Interface Protocols, including metadata and operational info (e.g., licensing or consenting) for their exchange.
3. A policy area that promotes and educates the responsible sharing through actions on public education, proper investments for workforce skills. The experts discussed and agreed that, although Europe has numerous strengths due to its public healthcare systems, strong expertise in genomics and infrastructures, there are some weaknesses and threats to address, like the fragmentation of skills including the AI and Big Data skills, the human resources capacity and significant translational complexity to assure socio-economic benefits.
4. A policy area that protects and secures these data. The discussions highlighted that, even within a well-designed legal framework regulating the proper use of genomics information built on dialog with and trust of the citizens, genomics data is, like all personal and health-related data, a target of interest of organised crime. Security, and the protection against breaches and unauthorised use, must be an

important component of the whole framework as there are cases where identification based on genomic data is feasible. It is important to rethink how data is protected in this context, and how technologies are being developed such as: traditional encryption (which can protect the data when stored or in transit, but not when being used in computations); homomorphic encryption (that allows computation on encrypted data); blockchain and related technologies, that don't protect privacy but allow strong accountability and traceability, for example when logging operations in a hospital system. There are other interesting approaches like 'pseudonymisation', which can enable safe linkage of molecular and clinical data.

3.2 Better economy

The economic angle was addressed by focusing on:

- economic impact assessment, intended as calculation of costs of adopting a particular technology, intervention or policy and recognition of their anticipated benefits; this should also include societal impact, as the impact of genomics technologies goes beyond profits for technology proprietaries and investors;
- funding, as in the provision of financial resources to finance projects, actions, programmes or policies on genomics.

Personalised medicine (as defined by ICPerMed) puts the single individual at the heart of the healthcare process and aims to achieve the most optimal outcome at the individual level (patient and citizen). For such a change in medicine practice to become mainstream, solid health and genetics or omics research results are needed, but more than that, the cost effectiveness of the approaches need to be considered, possibly demonstrated and streamlining its use in practical and concrete terms is also needed. Given the tremendous technology advances during the past years, harnessing the transformative power of genetics and genomics for the benefit of patients does not appear as costly nowadays as it has been in the past.

For example, Mammaprint and other genetic tests have shown that traditional processes for assessing costs and cost-benefit analyses need to be adapted to the circumstances of personalised healthcare. Clinical trials have new designs and end-points and the speed at which decisions are taken on reimbursements and authorisations conditions the speed at which data is acquired and effectiveness or benefit demonstrated (or not). The genetics and the genomics fields have called for a shift towards evidence-generating medicine, towards the use of real world data to inform health technology assessment decisions. The views of stakeholders like patients, health care professionals, insurance companies and health technology assessment (HTA) organisations are important to inform these decisions. Communities of practice like those within ISPOR (International Society for Pharmacoeconomics and Outcomes Research), EUnetHTA (European Network for Health Technology Assessment), EMA (European Medicines Agency) or national HTA agencies can support the definition of revised protocols and processes that ensure more flexible assessments, fully transparent and fair that do not delay or limit access to improved screening, diagnostic, guided therapy and care.

Two important aspects of streamlining the use of genetics and genomics in practice are access to tests and eligibility criteria. Take the example of cancer predisposition gene testing such as BRCA testing presented at the conference; eligibility to BRCA1 and BRCA2 genetic testing, even in cancer patients is complex in most EU health systems. However, Nazneen Rahman presentation on BRCA1 and BRCA2 genetic testing suggested it does not have to be so – a simple criterion for eligibility would be to offer testing if the chance of the individual to carry the mutation is higher than 10%. This chance threshold corresponds to 6 very simple cases calculated from modelling studies and real world data: 1) individual has ovarian cancer 2) individual has breast cancer <= 45 years, 3) individual has had two primary breast cancers ≤60 years, 4) individual has Triple Negative Breast Cancer, 5) individual has Male Breast Cancer or 6) individual has Breast Cancer + a parent, a child or a sibling with any of the above criteria.

The beauty of such simplicity is that it also allows for an easy calculation of the estimated needs for testing on a regular basis ensuring funding and material stocks.

In this particular example, the total needs have been estimated at around 127,000 tests/year for 10 of the biggest European countries and the UK experience has shown that the use of these is feasible, welcomed by patients, oncologists and geneticists as well as cost effective. The underlying concept (e.g. individual is tested if s/he has a 10% (or another value) chance of carrying the mutation) can be applied to several other predisposition tests.

Genomics can be applied to screening (or population screening), improve risk prediction and prevention of disease, and benefit economic and social aspects as well. The example on breast cancer demonstrates this and although this is an area where the application is well-established, it is not unique and many other medical areas, like rare diseases, are equally well-established and will benefit from the same engagement.

Risk prediction is important for clinical management. In the case of breast cancer for example risk prediction models are crucial to determine whether a woman is at low, moderate or high risk, which then guides the clinical recommendations for prevention and screening, risk reduction medication and prophylactic surgery according to risk categories. Improving the predictive power of risk models through applications of genomics is an important step toward targeted screening and prevention. The main risk factors for breast cancer include biological risk factors (age, hormonal factors, mammographic breast density), lifestyle risk factors (reproductive history, alcohol consumption, smoking, physical activity, obesity, use of hormone replacement therapy (HRT), radiation exposure) and family history. Currently, high-risk women are primarily identified on the basis of family history and mutation screening of two genes (BRCA1 and BRCA2). This approach has the limitation of missing women without family history but with a significant genetic predisposition. Jacek Gronwald discussed the utility of BRCA1 testing for prophylaxis and targeted therapy and also detailed the contribution of mutations in other risk-associated alleles in genes such as PALB2 and CHEK2 to the burden of familial breast and ovarian cancer.

Discussions on population based screening for BRCA carriers or testing of other genes in the clinical setting are abundant in the clinical oncology setting. Some of the questions that still prevail are related to determining which genes are truly associated with the risk of breast cancer; determining robust risk estimates for these variants/genes; determining the subtype-specific risks associated with these genes and whether these genes confer risks to other cancers.

In addition to the high and moderate risk genes, which account for only a small proportion of breast cancer cases in the general population, multiple common breast cancer susceptibility variants have been identified through genome-wide association studies (GWAS). These variants confer small risks individually, but their combined effect, when summarized as a polygenic risk score (PRS), can be substantial and their use in risk prediction models such as the Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm (BOADICEA) can support better stratification of women according to their risk of developing breast cancer. Polygenic Risk Scores (PRS) for breast cancer and ovarian cancer are predictive of cancer risk: incorporating the PRS into computer-assisted systems (like BOADICEA) could significantly affect the risk score and shift the risk class for approximately 20% of individuals from these families. The predictive power of risk prediction models thus has major implications for personalised risk-based screening and for risk-reduction interventions.

In terms of funding, Horizon Europe (and in particular the Cancer Mission) is seen by the scientific community as an essential mechanism to facilitate translation of genetics and genomics to the clinical practice, in oncology. An inclusive strategy that considers the transition of research outputs to the next stage in the translational pipeline is essential. Projects should complement each other, and 'results' (and state of the art technology) must be fit for the next step in the innovation cycle and comply with the end user demand. Such a strategy could reduce costs of R&D, enable innovation, allow patient stratification for benefit and risk, provide faster safety and efficacy in high risk groups, optimise indications and accelerate access to medicines, new outcomes measures and effectiveness data, improving EMA and HTA decision making and the ability to define the impact of such decisions, optimising use of medicines through ongoing monitoring.

Last but not least, the debate highlighted the presence of yet another important interface between citizens and genomics - the expanding sector of 'recreational genetics'. Citizens are customers, pay for and share a wealth of information that can be accessed and used by biotech companies without a clear mechanism for sharing the profits (with the individual and with society at large). Regulatory frameworks are not well adapted to this 'social network' type of research, to a form of 'surveillance capitalism' where populations' (genomics) data is harvested by private or public institutions. It is important to build a solid legal framework based on shared values and trust, where the interests of fully-informed citizens are centre stage.

3.3 Capitalise on existing 'infrastructures'

During the workshop, the word 'infrastructure' was used with different meanings and is considered under this chapter as any coordinated action (a project, facility, national or international organization, initiative, consortium or movement) that is fostering action, cooperation and coordination in this area at regional, national or international level.

The following list (in alphabetical order) is the result of our mapping of the infrastructures presented or mentioned during the workshop. Where possible, a URL is provided.

By browsing this list, it is quite evident that the areas of interest to this conference are well addressed in terms of 'infrastructure'. Nevertheless, while this admittedly non-exhaustive list of infrastructures is a start, a complete mapping is strongly needed for well-coordinated encompassing to joint efforts.

- BBMRI-ERIC. The Biobanks and BioMolecular Resources Infrastructure - European Research Infrastructure Consortium (http://www.bbmri-eric.eu/) is a European research infrastructure for biobanking. It brings together all the main players from the biobanking field (researchers, biobankers, industry, and patients) to boost biomedical research. Its goal is to make new treatments possible.
- BCAC. The Breast Cancer Association Consortium (http://bcac.ccge.medschl.cam.ac.uk/) is a forum of investigators interested in the inherited risk of breast cancer. The aim of the consortium is to combine data from many studies, and to provide a reliable assessment of the risks associated with these genes.
- BRIDGES. The BRIDGES project (https://bridges-research.eu/) aims to build a knowledge base that will allow identification of women at high-risk of breast cancer.
- Cancer Core Europe. Cancer Core Europe (https://www.cancercoreeurope.eu/) is a European cancer association created to construct a sustainable, high level, shared research infrastructure platform with research collaborations and taskforces (data sharing, clinical trials, genomics, immunotherapy, imaging, legal & ethical problems, and education & training).
- CIMBA. The Consortium of Investigators of modifiers of BRCA1/2 (http://cimba.ccge.medschl.cam.ac.uk/) was formed by a collaborative group of researchers working on genetic modifiers of cancer risk in BRCA1 and BRCA2 mutation carriers. The aim of CIMBA is to provide sufficient sample sizes to allow large scale studies in order to evaluate reliably the effects of genetic modifiers.
- CINECA. The Common Infrastructure for National Cohorts in Europe, Canada, and Africa (https://www.cineca-project.eu/) is a consortium established to create one of the largest cross-continental implementations of human genetic and phenotypic data federation and interoperability with a focus on common (complex) disease, one of the world's most significant health burdens.
- CORBEL. The Coordinated Research Infrastructures Building Enduring Life-science Services (https://www.corbel-project.eu) is an initiative of thirteen new biological and medical research infrastructures (BMS RIs), which together aims to create a platform for harmonised user access to biological and medical technologies, biological samples and data services required by cutting-edge biomedical research.
- DKTK. The German Cancer Consortium (https://dktk.dkfz.de) is a long-term, joint initiative involving the German Federal Ministry of Education and Research (BMBF), participating German states and the DKFZ. The main aim of the DKTK is to discover, develop, test and apply new personalized oncology strategies.
- DPPH. The Data Protection in Personalized Health (https://dpph.ch/) is a Swiss project funded by the Strategic Focus Area Personalized Health and Related Technologies (PHRT) of the ETH Board. It seeks to address the main scalability, privacy, security and ethical challenges of data sharing for enabling effective P4 medicine (Predictive, Preventive, Personalized and Participator), by defining an optimal balance between usability, scalability and data protection, and deploying an appropriate set of computing tools to make it happen.
- ECCO. The European CanCer Organisation (https://www.ecco-org.eu) is a multidisciplinary organisation that connects and responds to all stakeholders in oncology Europe-wide. It provides a cohesive platform for European cancer societies and organisations to work together to improve cancer patient outcomes and to be the unified voice of the European cancer professionals' community when addressing common policy issues.
- ECSEL JU. The ECSEL Joint Undertaking (https://www.ecsel.eu) is the Public-Private Partnership for Electronic Components and Systems. It funds Research, Development and Innovation projects for world-class expertise in these key enabling technologies, essential for Europe's competitive leadership in the era of the digital economy. Through the ECSEL JU, the European industry, SMEs and Research and Technology Organisations are supported and co-financed by 30 ECSEL Participating States and the European Union. ECSEL JU established Lighthouse Initiatives (a concept introduced by ECSEL JU to

signpost specific subjects of common European interest): one of them is 'Health.E', aimed to accelerate the innovation in medical electronic systems.

- EIT. The European Institute of Innovation and Technology (https://eit.europa.eu/) is an EU body created by the European Union in 2008 to strengthen Europe's ability to innovate. The EIT is an integral part of Horizon 2020, the EU's Framework Programme for Research and Innovation.
- ELIXIR. ELIXIR (https://elixir-europe.org) is an intergovernmental organisation that brings together life science resources from across Europe. These resources include databases, software tools, training materials, cloud storage and supercomputers. The goal of ELIXIR is to coordinate these resources so that they form a single infrastructure. This infrastructure makes it easier for scientists to find and share data, exchange expertise, and agree on best practices.
- EMBL-EBI. The European Molecular Biology Laboratory (EMBL, https://www.embl.de) is Europe's flagship laboratory for the life sciences – an intergovernmental organisation with more than 80 independent research groups covering the spectrum of molecular biology. The European Institute of Bioinformatics (EBI, https://www.ebi.ac.uk) is part of the family and on biological data services, research and training in bioinformatics. This intergovernmental research organisation is a trusted data provider for life sciences and hosts a number of most valuable publicly open and free to use life science resources, including biomedical databases, analysis tools and bio-ontologies.
- ENCR. The European Network of Cancer Registries (https://www.encr.eu/), established within the framework of the Europe Against Cancer Programme of the European Commission, has been in operation since 1990. It promotes collaboration between cancer registries, defines data collection standards, provides training for cancer registry personnel and regularly disseminates information on incidence and mortality from cancer in the European Union and Europe.
- ENIGMA. The Evidence-based Network for the Interpretation of Germline Mutant Alleles (https://enigmaconsortium.org/) is an international consortium of investigators focused on determining the clinical significance of sequence variants in BRCA1, BRCA2 and other known or suspected breast cancer genes, to provide this expert opinion to global database and classification initiatives, and to explore optimal avenues of communication of such information at the provider and patient level.
- EOSC-Life. The European Open Science Cloud (EOSC) for Life (http://www.eosc-life.eu/) aims to bring the capabilities of big science projects to the wider research community. Its goal is make sure that life-scientists can find, access and integrate life-science data for analysis and reuse in academic and industrial research.
- ERAPERMED. ERA PerMed (http://www.erapermed.eu/) is an ERA-Net Cofund, supported by 32 partners from 23 countries and co-funded by the European Commission. To align national research strategies, promote excellence, reinforce the competitiveness of European players in personalised medicine, and enhance the European collaboration with non-EU countries, national funding organisations have agreed to launch Joint Transnational Calls for collaborative innovative research projects in personalised medicine.
- EUnetHTA. The European network for health technology assessment (https://www.eunethta.eu/) is a network, established to create an effective and sustainable network for health technology assessment (HTA) across Europe that could develop and implement practical tools to provide reliable, timely, transparent and transferable information to contribute to HTAs in Members States.
- Europa Donna. Europa Donna - The European Breast Cancer Coalition (https://www.europadonna.org/) is an independent non-profit organisation whose members are affiliated groups from countries throughout Europe. Europa Donna works to raise awareness of breast cancer and to improve breast cancer services by promoting early detection, optimal treatment and research.
- European Alliance for Personalised Medicine. The European Alliance for Personalised Medicine (https://www.euapm.eu/) was established with the aim of improving patient care by accelerating the development, delivery and uptake of personalised medicine and earlier diagnostics, through consensus.
- GA4GH. The Global Alliance for Genomics and Health (https:/www.ga4gh.org/) is an international, non-profit alliance formed in 2013 aimed at accelerating the potential of research and medicine towards the advancement of human health. Bringing together 500+ leading organizations working in healthcare, research, patient advocacy, life science, and information technology, the GA4GH community

is working together to create frameworks and standards to enable the responsible, voluntary, and secure sharing of genomic and health-related data.

- GENIE. The Genomics Evidence Neoplasia Information Exchange (https://sagebionetworks.org/research-projects/aacr-project-genie/) is a multi-phase, multi-year, national and international project that catalyzes precision oncology through the development of a regulatory-grade registry aggregating and linking clinical-grade cancer genomic data with clinical outcomes from tens of thousands of cancer patients treated at participating institutions.
- GENOME Canada. Genome Canada (https://www.genomecanada.ca) is a not-for-profit organization, funded by the Government of Canada, aimed to act as a catalyst for developing and applying genomics and genomic-based technologies to create economic and social benefits for Canadians.
- Genomics England. Genomics England (https://www.genomicsengland.co.uk), with the consent of participants and the support of the public, is creating a lasting legacy for patients, the NHS and the UK economy, through the sequencing of 100,000 genomes. It was set up to deliver the 100,000 Genomes Project, the flagship project to sequence 100,000 whole genomes from National Health Service (NHS) patients with rare diseases, and their families, as well as patients with common cancers. This project is mainly funded by the National Institute for Health Research and NHS England. The Wellcome Trust, Cancer Research UK and the Medical Research Council have also funded research and infrastructure in the programme.
- Global Genomic Medicine Collaborative. The Global Genomic Medicine collaborative (https://g2mc.org) is an organization that is creating a community of global leaders dedicated to advancing genomic medicine implementation in clinical care.
- GO FAIR. GO FAIR (https://www.go-fair.org) is a bottom-up, stakeholder-driven and self-governed initiative that aims to implement the FAIR data principles, making data Findable, Accessible, Interoperable and Reusable. It offers an open and inclusive ecosystem for individuals, institutions and organisations working together through Implementation Networks (INs). The INs are active in three activity pillars: GO CHANGE (focussed on focus on priorities, policies and incentives for implementing FAIR), GO TRAIN, (on FAIR awareness and skills development training.) and GO BUILD (focussed on FAIR technology).
- HGVS. The Human Genome Variation Society (https://www.hgvs.org) aims to foster discovery and characterization of genomic variations including population distribution and phenotypic associations. They promote collection, documentation and free distribution of genomic variation information and associated clinical variations and endeavour to foster the development of the necessary methodology and informatics.
- HUGO. The Human Genome Organisation (http://www.hugo-international.org) is an organization involved in the Human Genome Project, a project about mapping the human genome. It seeks to bring the benefits of Genomic Sciences to Humanity by promoting fundamental genomic research within nations and throughout the world, by fostering scientific exchange in genomics with a particular emphasis on scientifically developing and emerging countries, by supporting discourse in the ethics of genetics and genomics with a global perspective
- ICGC. The International Cancer Genome Consortium (https://icgc.org) is a voluntary scientific organization that provides a forum for collaboration among the world's leading cancer and genomic researchers. The ICGC was launched in 2008 to coordinate large-scale cancer genome studies in tumours from 50 cancer types and/or subtypes that are of main importance across the globe. It incorporates data from The Cancer Genome Atlas (TCGA) and the Sanger Cancer Genome Project.
- Inspire2Live. Inspire2Live (https://inspire2live.org/) is an international movement founded by the people who also started charity events like Alpe d'HuZes, currently Holland's biggest charity action (30M Euros) and many others, such as the Belgian Ven2-4Cancer.
- IRDiRC. The International Rare Disease Research Consortium (http://www.irdirc.org/) aims to enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.
- OECI. The European Organization of Cancer Institutes (https://www.oeci.eu/) is a non-governmental, no-profit legal Entity established in 1979 to promote greater cooperation among European Cancer Centres

and Institutes. OECI is a network presently regrouping 93 members collaborating to reduce fragmentation and to give to all European cancer patients the possibility of receiving the best available care.

- PCAWG. The PanCancer Analysis of Whole Genomes is an international collaboration to identify common patterns of mutation in more than 2,800 cancer whole genomes from the International Cancer Genome Consortium.
- RIPATHS. The Research Infrastructure imPact Assessment PaTHwayS (https://ri-paths.eu/) aims to develop a framework describing the socio-economic impact of research infrastructures (RIs) and their related financial investments.
- The DECIPHER Community. The DECIPHER Community is an international community of academic departments of clinical genetics and rare disease genomics that contribute to the DECIPHER (DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources, https://decipher.sanger.ac.uk).
- The Human Variome Project. The Human Variome Project (http://www.humanvariomeproject.org/) is an international non-governmental organisation that is working to ensure that all information on genetic variation and its effect on human health can be collected, curated, interpreted and shared freely and openly.
- WFPHA. The World Federation of Public Health Associations (https://www.wfpha.org) is an international, nongovernmental organization composed of over 115 associations member, mostly multidisciplinary national public health associations, and representing around 5 million public health professionals worldwide. Its mission is to promote and protect global public health.

3.4 Develop educational programmes

Education and training was a topic repeatedly mentioned in many of the conference sessions. Keywords were:

- multidisciplinary, i.e. building on, and targeting different academic disciplines and professions that work together in Genomics;
- professional, the creation of specialised training and life-long learning in professional schools through which participants acquire content knowledge and learn to apply novel genomic techniques;
- non-professional, to increase understanding of Genomics by all and allow citizens to engage in 'public participation in Science'.

For example:

- Going back to a 'common understanding' for genomic data interoperability, it is easy to understand the need for a multidisciplinary approach, that can be defined and realised through the setup of multidisciplinary teams of experts under a well-defined project governance. Such governance should oversee at least four 'sets' of policy areas, one being responsible sharing through actions on public education, investments for workforce skills, information and communications technology infrastructures for life sciences, and trustworthy computational-based analyses.
- As for well-informed, educated citizens, the examples that surface from the surveillance capitalism discussions illustrate the need for a regulatory framework to address the use of citizens' (genomics) data including ensuring consumers are provided the information needed and are educated to take mindful decisions regarding the use of their own data (and in this context, their genomic information, that is also that of their immediate relatives).

3.5 Effective regulation

A significant part of the regulation discussions was centred on the GDPR, but participants debated many other regulatory and policy needs stemming from the links and interactions between genomics, genomic research, innovation and society. One case in point was regulation and innovation. 'What is innovation?' and 'What is the

regulatory framework needed to translate health innovation into healthcare systems in the EU in a smooth and quick manner?'

At the conference, 'innovation' was presented as the 'translation of knowledge and insight into value'. In the case of healthcare, value to patients and society can change over time and is seen as a 'moving target'. There are three reasons for this:

1. technology evolves very fast in this sector and comparisons in terms of improved outcomes with previous technologies are inevitable;
2. the main targets, i.e. clinicians and patients, are fast adopters of new technology and use it more efficiently and effectively paralleled by higher expectations;
3. prices are dropping quickly and there is a fast appearance (not just a simple turn-over) of cheaper relevant alternative new technologies.

To approach such a moving target, a self-learning healthcare system based on the concept of evidence-generating care is an obvious (albeit not simple) approach to quickly capture value. Evidence-generating medicine comes however with its own challenges, which include:

- learn by doing;
- acceptance by both doctors and patients – evidence-based medicine is the prevailing practice and there are good reasons that it is so;
- potential costs and failure;
- empowerment of citizens to manage their own health.

Like for any other technology, the value of genomics can, and must be, shaped by payers and health care providers. There is no doubt that tailored, local solutions are needed to address barriers specific to each country. Before regulations are considered, the identification and development of such solutions should be supported by inclusive EU-level debates and initiatives that will shape the regulations themselves as well.

Coordination (including at EU-level) is needed considering the multiple regulatory initiatives, the changing science, the challenge of 'big' biological data (with its four 'big data' dimensions: volume, variety, velocity and veracity) as well as a potential shift in the reimbursements' landscape.

A good example of such coordination is the development of a policy framework for HTA: since January 2018, more than 80 organisations from 29 countries groups are working together on this issue in the EUnetHTA joint action initiative (https://eunethta.eu/). This and other actions, like the ECSEL Joint Undertaking Health.E lighthouse initiative on health (https://www.ecsel.eu/healthe), are expected to be strictly in contact as their efforts aims to tackle similar challenges.

Another example relates to the European '1+ Million Genomes' Initiative that aims to:

- integrate genomic data into electronic health records and use this to support routine clinical decisions and an evidence-based and sustainable health system;
- engage citizens by supporting their engagement in the use of genomic data for their own health and ability to contribute to research for societal benefit;
- foster collaborative multi-disciplinary research in a hybrid research/care model to address the areas of most clinical need and ensure system adoption;
- support a precision medicine industry encompassing development of genomics, health and technology enterprises in each country as well as encourage investment of leading global pharma, biotechnology and technology companies.

At the policy level though, how do regulators plan to coordinate and support European national strategies and initiatives that can fulfil their expected aims and benefits and abide by their principles? The following are some points for reflection for this initiative to live to its high expectations:

- leverage and maximise the use of existing resources;
- develop a federated 'knowledge-base' of genomic and health information;

- surround these 'knowledge-base' with platforms for clinical discussions, training, research, innovation & entrepreneurship;
- establish a cross-border network of expertise;
- create a platform for participant engagement;
- promote transfer of technology and its application via 'instruments' designed by the Commission or Member States';
- fund impactful innovation through public private partnerships.

3.6 Forging ties with society

As discussed during the conference, Society in this context encompasses and focuses on the following subjects:

- individual, i.e. as single human being with human rights;
- citizen, i.e. an individual who is a member of a particular country and has rights because of being born there or because of being given rights, including right to health;
- patient, i.e. as person receiving or registered to receive medical treatment.

If done right, the application of genomics to personalised medicine will improve many aspects of public health: precision diagnostics (e.g. for rare diseases), precision treatment (e.g. cancer therapies), personalised prevention (e.g. against common diseases) and importantly pharmacogenomics (i.e. anticipate drug responses and tailored treatments). Efficiently integrating genomics into the public healthcare systems has the potential to produce a virtuous cycle of 'evidence-generating medicine' where new patients, samples, data and associated health outcomes improve the existing knowledge.

Citizens need to be engaged, as any citizen is a potential future patient or will be interested in the health outcome of a relative or a friend. Focus groups and surveys are important tools to engage with the citizens, for example on personalised medicine. Different 'risk' levels or what the concept of 'intermediate risk' means are complex concepts that need to be clarified in this context, for example. Citizens' health and well-being should be the heart of any academic or private research activity in this area but citizens are often the group with the least influence on all other stakeholders. Citizen engagement in such public health discussions needs to consider both the 'healthy citizen' and the 'patient' as well as the 'survivor.' There is a political will for inclusion but the citizen appears to feel excluded.

Citizen engagement is particularly relevant to ensure 'responsible sharing' as discussed previously. As individuals, the efforts to share human genome sequences or data from health records should be framed within the Universal Declaration of Human Rights, in particular Article 27: 'Everyone has the right freely to participate in the cultural life of the community, to enjoy the arts and to share in scientific advancement and its benefits.' This declaration is legally actionable, and Member States should use it to establish mechanisms for collaborative, open spaces and infrastructures for science to thrive.

By using the term 'everyone', article 27 of the Universal Declaration of Human Rights recalls another concept discussed at the conference that is equity. Genomics, but more in general 'digital health' as electronic forms of health communications, can potentially help reduce social inequities but needs to be properly framed. Digital health can be an instrument for improving social 'equity' and a channel to:

- ensure diversity when performing research (e.g. current Genome Wide Association Studies are overwhelmingly focused in white European cohorts);
- provide distributive justice, to ensure that all? Individuals and groups fairly benefit from the health benefits of personalised medicine;
- facilitate proper balance between Individual Benefits (privacy) and Collective Benefits (sharing);
- balance social imbalances (like informed consent and return of results vs levels of literacy) and reduce regional disparities (like rural vs urban areas);
- foster benefits through an efficient reciprocity between health systems and the citizens (contribution of information as a patient to improve databases, resulting in better healthcare and screening).

A concrete example that would benefit from more coordinated citizen engagement is biobanking; biobanking is crucial in all aspects of human genome sequencing and personalised medicine, including all phases of medicine development and diagnostics. Biobanking is built on trust from the citizens that agree to deposit and share their biological material and can only work if that trust is well managed and fully respected.

Large biobank projects (like BBMRI-ERIC or the Estonian Biobank presented at the conference) need to integrate citizens in their advisory boards to support decisions related to ethical, legal and societal challenges linked to the samples and their use for research. A biobank should be prospective, longitudinal and volunteer-based, and needs clear access and consent rules to be open for scientific research. Information can be returned to the citizens (donors) who agree to receive it, in an internationally agreed form (like 'polygenetic risk score'), which requires professional counselling to be interpreted correctly (see below). Some important needs listed in relation to biobanking are:

- the need for improving the utilization of existing biosamples (currently, only about 10% are being used for research);
- the need of developing a code of conduct to elaborate biobanking policies compliant with the EU GDPR. This also includes the need for an exchange between patients and policy makers so that any particularities linked to genomics data may eventually be reflected on the regulation;
- the need for implementing strategies to check and validate the in depth understanding of the consent form by donors (how well and correctly 'informed' a donor really is);
- the need for meeting citizen's expectations on the return of research results, which are constantly evolving;
- the need for avoiding duplication and fragmentation across different biobanking initiatives.

If the case of biobanking was an example of how the scientific community can benefit from citizen engagement, from the citizens point of view there are other questions that were addressed at the conference, especially in relation to the moment in which they become patients: What's wrong? (Diagnosis); What's the future? (Prognosis); What's to be done? (Therapy); Why did it happen? (Aetiology); Will it happen again? (Recurrence risk); Will it be as bad? (Clinical burden).

Given the technology advances of the past years and the substantial decrease in the cost of sequencing a genome (which might reach $100 within the next 5 years), it is now cheap enough to harness the transformative power of genomics for the benefit of patients. For example, a large body of evidence supporting the clinical utility is accumulating with a consistent 20 to 30% yield of diagnosis for suspected rare diseases. This means that for 20-30% more patients the 'diagnostic odyssey' ends, a painful, emotionally draining and costly journey – not to mention that a diagnosis can also finally mean access to cure and treatment. In this context, it is fundamental to improve professional counselling, especially in genetics. Currently, genetic counselling aims at detecting and excluding the presence of high-risk alleles. More inclusive and effective actions could aim at assessing individual risks using genetic information for common and rare variants as well as for other modifiable risk factors. Non-genetic risk factors should be incorporated into risk prediction and modifiable risk factors should be taken into consideration. Anyone could be tested and the results would likely be a continuum or risk scores rather than 'positive' or 'negative' results. Thus, it would be possible to identify a larger group of individuals at high risk of cancer with much greater precision than it is currently possible, allowing a more effective use of available resources to those who would benefit the most from screening and preventive strategies.

Improving of professional counselling, especially the genetic one, needs to be 'citizen-centric'. In the Genomics England Project an example of such a counselling described at the conference was the Genomics England Project. One of the aims of the project is to communicate the information regarding the patient's main condition for which they were referred to all participants who agree to receive it. For example, Participants can opt in to receive feedback on a selection of known genetic alterations of high clinical significance and non-affected parents of children with rare disease can also chose to be informed about their carrier status. The project also promotes ethics and transparency values and ensures that the interests of participants are at the centre. In this regard,a broad consent for translational research has been obtained including detailed data access procedures. The project also foresees a new service to enable volunteers to pay for a personalised report of their genetic makeup. With the permission of these volunteers, the genetic data will be made available to researchers and scientists; volunteers will work with NHS and patient groups to lead the development of the service. Sharing the data is a form of 'genetic solidarity and altruism', important values of the project and, gives rise to opportunities to help others, perfectly fitting with article 27 of the Universal Declaration of Human Rights.

List of abbreviations and definitions

AI	Artificial Intelligence
API	Application Programming Interface
ATM	Ataxia-Telangiesctasia Mutated gene
BBMRI	Biobanks and BioMolecular Resources Infrastructure
BBMRI-ERIC	Biobanks and BioMolecular Resources Infrastructure - European Research Infrastructure Consortium
BCAC	Breast Cancer Association Consortium
BOADICEA	Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm
BRCA1	BReast CAncer type 1 susceptibility protein
BRCA2	BReast CAncer type 2 susceptibility protein
BRIDGES	BReast Cancer Risk after Diagnostic Gene Sequencing
CHEK2	CHEckpoint Kinase 2
CIMBA	Consortium of Investigators of Modifiers of BRCA1/2
CINECA	Common Infrastructure for National Cohorts in Europe, Canada, and Africa
CORBEL	Coordinated Research Infrastructures Building Enduring Life-science Services
CPG	Cancer Predisposition Gene
DECIPHER	DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources
DG CNECT	Commission's Directorate-General for Communications Networks, Content and Technology
DG RTD	Commission's Directorate-General for Research and Innovation
DG SANTE	Commission's Directorate-General for Health and Food Safety
DKTK	Deutschen Konsortium für Translationale Krebsforschung - German Cancer Consortium
DNA	DeoxyriboNucleic Acid
DOIP	Digital Object Interface Protocol
DPPH	Data Protection in Personalized Health
EC	European Commission
ECCO	European CanCer Organisation
ECIBC	European Commission Initiative on Breast Cancer
ECSEL JU	Electronic Components and Systems for European Leadership Joint Undertaking
EIT	European Institute of Innovation and Technology
EIT Health	Health knowledge and innovation community of the European Institute of Innovation and Technology
ELIXIR	European Life-Science Infrastructure for Biological Information
EMA	European Medicines Agency
EMBL	European Molecular Biology Laboratory
EMBL-EBI	European Molecular Biology Laboratory - European Bioinformatics Institute
ENCR	European Network of Cancer Registries
ENIGMA	Evidence-based Network for the Interpretation of Germline Mutant Alleles
EOSC	European Open Science Cloud
EPFL	Ecole Polytechnique Fédérale de Lausanne

ETH	Eidgenössische Technische Hochschule of Zurich
EU	European Union
EUnetHTA	European Network for Health Technology Assessment
FAIR	Findable, Accessible, Interoperable, Reusable
GA4GH	Global Alliance for Genomics and Health
GDRP	General Data Protection Regulation
GENIE	Genomics Evidence Neoplasia Information Exchange
GWAS	Genome-Wide Association Study
HGVS	Human Genome Variation Society
HRT	Hormone Replacement Therapy
HTA	Health Technology Assessment
HUGO	Human Genome Organisation
ICGC	International Cancer Genome Consortium
ICPerMed	International Consortium for Personalised Medicine
IRDiRC	International Rare Disease Research Consortium
ISPOR	International Society for Pharmacoeconomics and Outcomes Research
IT	Information Technology
JRC	Joint Research Centre
LUMC	Leiden University Medical Center
MEGA	Million European Genomes Alliance
MRI	Magnetic Resonance Imaging
MS	Member States
NCT	National Center for Tumor Diseases
NHS	United Kingdom National Health System
NIJZ	National Institute of Public Health of Slovenia
OECI	Organisation of European Cancer Institutes
PALB2	Partner and Localizer of BRCA2
PCAWG	Pan-Cancer Analysis of Whole Genomes
PHRT	Personalized Health and Related Technologies
PMU	Pomeranian Medical University of Poland
PRS	Polygenic Risk Scores
RIPATHS	Research Infrastructure imPact Assessment PaTHwayS
RNA	RiboNucleic Acid
R&D	Research and Development
SME	Small and Medium Enterprises
UCSF	University of California San Francisco
VHIO	Vall d'Hebron Institute of Oncology
WFPHA	World Federation of Public Health Associations

Annex: Conference programme

European Commission

Integrating genomics into personalised healthcare: a science-for-policy perspective

My genome: our future

CONFERENCE PROGRAMME

12-13 February 2019
Brussels, Belgium

#mygenomeourfuture

Joint Research Centre

Contact information
Name: Guy Van den Eede
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EU Science Hub
https://ec.europa.eu/jrc

Integrating genomics into personalised healthcare: a science-for-policy perspective
My genome: our future
Conference Programme
12-13 February 2019
Brussels, Belgium

Scientific Steering Committee Members

Tit Albreht
National Institute of Public Health (NIJZ), Slovenia

Rolf Apweiler
The European Molecular Biology Laboratory - European Bioinformatics Institute (EMBL-EBI), United Kingdom

Mauro Giacca
King's College London, United Kingdom

Peter Goodhand
Ontario Institute for Cancer Research, Canada

Jan Korbel
European Molecular Biology Laboratory (EMBL), Germany

Jacques Simard
Université Laval, Canada

Rita Schmutzler
Uniklinik Köln, Germany

Giorgio Stanta
University of Trieste, Italy

Marc Van den Bulcke
Sciensano, Belgium

Joris Vermeesch
University of Leuven, Belgium

Julia Wilson
Wellcome Trust Sanger Institute, United Kingdom

Local Steering Committee Members

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European Commission, Joint Research Centre, Italy

Sandra Caldeira
European Commission, Joint Research Centre, Italy

Ciarán Nicholl
European Commission, Joint Research Centre, Italy

Mauro Petrillo
European Commission, Joint Research Centre, Italy

Maddalena Querci
European Commission, Joint Research Centre, Italy

Guy Van den Eede
European Commission, Joint Research Centre, Belgium

Tuesday 12 February 2019

AM

8:00 - 9:15 Participants registration and welcome coffee

9:30 - 10:00 **Institutional addresses**

Chair: Vladimír Šucha
Director-General, European Commission, Directorate-General Joint Research Centre, Belgium

Martin Seychell
Deputy Director-General, European Commission, Directorate-General for Health and Food Safety, Belgium

Irene Norstedt
Acting Director, European Commission, Directorate-General for Communications Networks, Content and Technology, Belgium

10:00 - 13:15 Session I: Setting the stage

Chair: Joris Vermeesch
University of Leuven, Belgium

Co-chair: Marco Marsella
European Commission, Directorate-General for Communications Networks, Content and Technology, Luxembourg

Rapporteur: Jacques Simard
Université Laval, Canada

10:00 - 11:00 **Keynote presentations**

10:00 - 10:30 **Big data in healthcare and biology, opportunities and challenges**
Ewan Birney
European Molecular Biology Laboratory - European Bioinformatics Institute (EMBL-EBI), United Kingdom

10:30 - 11:00 **From clinical genetics to genomic medicine: a brief history**
Sir John Burn
Newcastle University, United Kingdom

11:00 - 11:30 Coffee break

11:30 - 13:15 **Moderated panel discussion with**

Peter Devilee
Leiden University Medical Center, The Netherlands

Mark Bale
Genomics England, United Kingdom

Denis Horgan
European Alliance for Personalised Medicine, Belgium

Peter Kapitein
Inspire2live, The Netherlands

13:15 - 14:15 Lunch break

PM

Tuesday 12 February 2019

14:15 - 14:45 Flash talks by young health innovators

Chair: Mauro Giacca
King's College London, United Kingdom

14:15 - 14:30 **Reinventing ADCs - Catalyzing the discovery and development of next-generation Antibody Drug Conjugates (ADCs) for targeted therapies**
Dominik Schumacher
Tubulis Technologies, Germany

14:30 - 14:45 **From genetics to metabolism, a translational effort towards cancer diagnostics**
Francesco Gatto
Elypta, Sweden

14:45 - 18:00 Session II: Genomics - opportunities and challenges

Chair: Giorgio Stanta
University of Trieste, Italy

Co-chair: Irene Norstedt
European Commission, Directorate-General for Research and Innovation, Belgium

Rapporteur: Rolf Apweiler
European Molecular Biology Laboratory - European Bioinformatics Institute (EMBL-EBI), United Kingdom

14:45 - 15:45 **Keynote presentations**

14:45 - 15:15 **Data sharing in genomic medicine: opportunities and challenges**
Jan Korbel
European Molecular Biology Laboratory (EMBL), Germany

15:15 - 15:45 **The internet for social machines**
Barend Mons
Leiden University Medical Center (LUMC), The Netherlands

15:45 - 16:15 Break

16:15 - 18:00 **Moderated panel discussion with**

Rodrigo Dienstmann
Vall d'Hebron Institute of Oncology (VHIO), Spain

Stefan Fröhling
National Center for Tumor Diseases (NCT), Germany

Serena Scollen
European Life-Science Infrastructure For Biological Information (ELIXIR), United Kingdom

18:00 End of day 1

18:00 - 19:00 Refreshments

Wednesday 13 February 2019

AM

8:00 - 8:45 Participants registration

8:45 - 12:00 Session III: The public health perspective

Chair: Tit Albreht
National Institute of Public Health (NIJZ), Slovenia

Co-chair: Elke Anklam
European Commission, Directorate-General Joint Research Centre, Belgium

Rapporteur: Marc Van den Bulcke
Sciensano, Belgium

8:45 - 9:45 **Keynote presentation**

The future of oncology research in Horizon Europe
Thierry Philip
Institut Curie and Organisation of European Cancer Institutes (OECI), France

9:45 - 10:15 Coffee break

10:15 - 12:00 **Moderated panel discussion with**

Nazneen Rahman
Independent healthcare consultant, United Kingdom

Jacek Gronwald
Pomeranian Medical University (PMU), Poland

Jan-Ingvar Jönsson
Swedish Research Council, Sweden

Laura van't Veer
University of California San Francisco (UCSF), Helen Diller Family Comprehensive Cancer Center, USA

12:00 - 13:00 Lunch break

13:00 - 13:30 Flash talks: A focus on genomics for rare diseases

Chair: Julia Wilson
Wellcome Trust Sanger Institute, United Kingdom

13:00 - 13:15 **SOLVE-RD: a diagnosis for every rare disease patient**
Han Brunner
Radboud University Medical Center (Radboudumc), The Netherlands

13:15 - 13:30 **European joint programme on rare diseases – bringing genomic tools and discoveries to clinics**
Daria Julkowska
National Institute of Health and Medical Research (INSERM), France

PM

Wednesday 13 February 2019

13:30 - 16:45 Session IV: The citizen and patient perspective

Chair: Peter Goodhand
Global Alliance for Genomics and Health, Canada

Co-chair: Ioana-Maria Gligor
European Commission, Directorate-General for Health and Food Safety, Belgium

Rapporteur: Rita Schmutzler
Uniklinik Köln, Germany

13:30 - 14:30 **Keynote presentations**

13:30 - 14:00 **Citizen and patient perspectives: The Quebec experience**
Bartha Knoppers
McGill University, Canada

14:00 - 14:30 **How biobanking can help deliver on the promises of personalised medicine**
Francesco Florindi
Biobanking and Biomolecular Resources Research Infrastructure - European Research Infrastructure Consortium (BBMRI-ERIC), Austria

14:30 - 15:00 Break

15:00 - 16:45 **Moderated panel discussion with**

Jane Kaye
Centre for Law, Health and Emerging Technologies (HeLEX), University of Oxford, United Kingdom

Bettina Borisch
University of Geneva, Switzerland

Jean-Pierre Hubaux
Ecole Polytechnique Fédérale de Lausanne (EPFL), Switzerland

Andres Metspalu
University of Tartu, Institute of Genomics, Estonia

16:45 - 17:30 Final keynote presentation

Chair: Guy Van den Eede
European Commission, Directorate-General for Health and Food Safety, Belgium

Final keynote presentation:
Genomic medicine programs of the National Human Genome Research Institute
Teri Manolio
National Human Genome Research Institute, National Institutes of Health (NIH), USA

17:30 Closing of the conference

Institutional addresses

Chair

Vladimír Šucha

Director-General, European Commission, Directorate-General Joint Research Centre, Belgium

Vladimir Šucha is Director-General of the Joint Research Centre of the European Commission, its in-house scientific service. He was Deputy Director-General of the JRC between 2012 and 2013. Prior to that, he spent 6 years serving as Director of Culture and Media in the Directorate-General for Education and Culture of the European Commission. Before joining the European Commission, he held various positions in the area of European and international affairs. Between 2005 and 2006, he was Director of the Slovak Research and Development Agency, the national body responsible for funding research. He was the principal advisor for European affairs to the minister of education of the Slovak Republic (2004-2005). He worked at the Slovak Representation to the EU in Brussels as research, education and culture counselor (2000-2004). In parallel, he has followed a long-term academic and research career, being a full professor in Slovakia and visiting professor/scientist at different academic institutions in many countries. He has published more than 100 scientific papers in peer reviewed journals.

Speaker

Martin Seychell

Deputy Director-General, European Commission, Directorate-General for Health and Food Safety, Belgium

Martin Seychell is Deputy Director-General of Health in the Health and Food Safety Directorate-General (SANTE) since 2014. He is a graduate in chemistry and pharmaceutical technology with specialization in chemical analysis. He has held various important positions on several government Boards and Commissions in Malta, including the Food Safety Commission and the Pesticides Board. Between 2001 and 2006, Mr Seychell occupied the post of Head of Directorate at the Malta Standards Authority. He has been responsible for the implementation of a number of EU directives in the areas of risk assessment, food safety, chemicals and cosmetic products legislation, and has actively participated in negotiations on major technical proposals such as the new chemicals legislation, REACH, and in screening processes in the areas of free movement of goods, environment and agriculture during the process leading to Malta's accession to the EU. Between 2006 and 2011, he held the post of Director of Environment in Malta and was responsible for a broad range of functions arising from the Maltese Environment Protection Act. In March 2011, he was appointed Deputy Director-General for Health and Consumers at the European Commission and was responsible for the Directorates dealing with Consumer affairs, Public health and Health systems and products (SANCO).

Speaker

Irene Norstedt

Acting Director, European Commission, Directorate-General for Communications Networks, Content and Technology,Belgium

Irene Norstedt is the Acting Director responsible for the Health Directorate of the DG for Research and Innovation at the European Commission. She is also Head of the Innovative and Personalised Medicine Unit. She has served at the European Commission since 1996 and was instrumental in the creation of the Innovative Medicines Initiative (IMI) in 2008. From 16 December 2014 to15 September 2015, Irene served as Acting Executive Director of the Innovative Medicines Initiative. Before joining the European Commission, she worked for the Swedish Life Science company, Biacore AB and at the Swedish Embassy in London. Irene studied biotechnology and polymer science, and holds a Master of Science (MSc) in Chemical Engineering.

Session I:

Setting the stage

Chair

Joris Vermeesch
Department Chair, University of Leuven, Belgium

The Vermeesch laboratory is focused on developing technologies for rare disease analysis, understanding the causes and mechanisms underlying rare developmental disorders and it primarily focuses on structural variation and mosaicism detection. The laboratory has been translating those technologies to leverage postnatal diagnosis, as well as preimplantation and prenatal to avoid transmission of disease alleles. The laboratory has also been focusing on embryonic development and early placentation and aims to unravel the mutational mechanisms active during those early developmental stages. Joris Vermeesch is the founder of Cartagenia, a start-up company specialized in genomic data analysis for clinical diagnostic laboratories, founder and coordinator of Genomics core Leuven and founder and president of the Leuven Institute of Genomics and Society (LIGAS). He has published over 300 papers and an H-index of 51.

Co-chair

Marco Marsella
Head of Unit, European Commission, Directorate-General for Communications Networks, Content and Technology, Luxembourg

Marco Marsella is Head of the "eHealth, Well-being, and Ageing" Unit of the Directorate General for Communications Networks, Content and Technology (DG CONNECT) of the European Commission. From 2016 to June 2018, Marco Marsella headed the Unit responsible for the Web Accessibility Directive, Safer Internet and Language Technologies. He has worked on policy development, innovation and research implementation in the areas of digital content, technologies for learning, e-inclusion and assistive technologies.

Rapporteur

Jacques Simard
Vice-Dean of Research and Graduate Studies, Université Laval, Canada

Jacques Simard holds a Canadian Research Chair in Oncogenetics and is Vice-Dean of Research and Graduate Studies at the Faculty of Medicine of Université Laval. He was the lead investigator (Bartha M. Knoppers co-lead) of the project Personalized Risk Stratification for Prevention and Early Detection of Breast Cancer (PERSPECTIVE) (2013-2018), designed to develop the tools needed to implement a risk stratification approach that would target breast cancer screening in women most likely to develop the disease. He leads (Anna Chiarelli co-lead) PERSPECTIVE: Integration and Implementation (2018-2022) to further improve personalized risk assessment, to provide risk-based prevention and early detection of breast cancer, and to determine the optimal implementation approaches in the Canadian health system.

Keynote speaker

Ewan Birney

Director, European Molecular Biology Laboratory - European Bioinformatics Institute (EMBL-EBI), United Kingdom

Ewan Birney is Director of EMBL-EBI, together with Rolf Apweiler, and runs a small research group. He is also EMBL-EBI's Joint Head of Research, alongside Nick Goldman. Ewan Birney led the analysis of the Human Genome gene set, mouse and chicken genomes and the ENCODE project, focusing on non-coding elements of the human genome. Ewan Birney's main areas of research include functional genomics, DNA algorithms, statistical methods to analyse genomic information (in particular, information associated with individual differences in humans and Medaka fish) and use of images for chromatin structure. Ewan Birney is a non-executive Director of Genomics England, and a consultant and advisor to a number of companies, including Oxford Nanopore Technologies, Dovetail Genomics and GSK. Ewan Birney was elected an EMBO member in 2012, a Fellow of the Royal Society in 2014 and a Fellow of the Academy of Medical Sciences in 2015.

Keynote speaker

Sir John Burn

Professor of Clinical Genetics, Newcastle University, United Kingdom

Sir John Burn has been a registered specialist in clinical genetics since 1982 and Professor since 1991. He helped construct, and led for two decades, the NHS genetic services in the Northern region of England. He helped conceive and brought to the frontier, the International Centre for Life to celebrate and advance genomics. He has been chair of the British Society of Human Genetics, a scientific advisor to the Department of Health, the Science and Technology Committee, the DDD project at the Sanger Centre and the UK 100,000 genome project. He has been a member of the Board of NHS England and now Chair of Newcastle Hospitals, responsible for one of the 7 genomic laboratory hubs. He helped create and currently Chair of QuantuMDx Ltd, a company developing point of care DNA testing devices suitable for low and middle-income countries.

Panellist

Peter Devilee

Head of Section, Leiden University Medical Center, The Netherlands

Peter Devilee has 35 years' experience in cancer genetics, including somatic genetics of tumors and the genetics of familial clustering of cancer, primarily breast and colorectal cancer, and paragangliomas. Since 2003, he is a professor of Tumor Genetics at the Leiden University Medical Center (Department of Human Genetics and the Department of Pathology). In the late nineties, he was the coordinator of the worldwide Breast Cancer Linkage Consortium, which made significant contributions to the understanding of the genetics of familial breast cancer. In 1995, he was co-discoverer of the BRCA2 gene, and in 2000, of the gene-defect underlying hereditary paragangliomas. His current works include searches for new cancer susceptibility and risk modifier genes in selected families and patient populations, studies directed towards understanding the polygenic nature of cancer susceptibility and the pathogenicity of unclassified variants in the BRCA1/2 genes. Since 2015, he has been coordinating the BRIDGES project, an EU-program aimed at developing a personalized breast cancer risk-estimation tool.

Panellist

Mark Bale

Head of Science Partnerships, Genomics England, United Kingdom

Mark Bale is the senior genomics policy expert at the Department of Health & Social Care and Head of Science Partnerships at Genomics England. He has had responsibility for several emerging areas of science and their ethical, legal and policy implications. The current emphasis is on genomics (particularly, the 100,000 Genomes Project) and emerging areas such as genome editing. At Genomics England, he coordinates the various partnerships within England, Scotland, Wales, and Northern Ireland, and links with other international partners. These include the EU 1 Million Genomes Initiative and representation at the Council of Europe and OECD.

Panellist

Denis Horgan

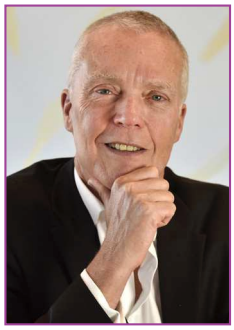
Executive Director, European Alliance for Personalised Medicine, Belgium

Denis Horgan is the Executive Director of the European Alliance for Personalised Medicine (EAPM). Horgan's background blends extensive expertise in health policy and issues advocacy with a unique understanding of how civil society interacts with today's political arena at the European and Member State level. Prior to working for the EAPM, he worked at the European Cancer Patient Coalition and in the European Parliament on a broad array of health issues relating to the pharmaceutical area and patient issues and for international NGOs on health development projects in Afghanistan, Mexico and Palestine. Throughout his career, Denis Horgan has developed numerous public affairs initiatives in the areas of advocacy, social marketing, policy support and launch, policy development and media relations. As a senior policy affairs manager, he has actively been involved in coalition building, grassroots advocacy, conferences development and development of policy platforms to support access to reimbursement/treatment at the political level and for institutions/ bodies. Specialities: European Union policy and legislation, government affairs, lobbying, patient relations, patients sector, communication, advocacy, NGO sector, Development sector.

Panellist

Peter Kapitein

Patient Advocate, Inspire2Live, The Netherlands



As a Patient Advocate of Inspire2Live, Peter Kapitein connects patients, researchers and clinicians to further research, treatments and care in The Netherlands, as well as globally. He organises congresses, lobbies the matrix of public authorities, health care organizations, insurance companies and health research institutes. Peter Kapitein also gives lectures and talks to help patients and society fight cancer, where possible and to live with it with a good quality of life. He writes blogs, articles and books that contribute to these topics. He has studied the Medical Industrial Complex, the complex in which the stakeholders in healthcare work together in a way that does not necessarily benefit the patient. Health care is (without bad intention) distracted from its essence, the patient. Peter Kapitein is the co-founder of Alpe d'HuZes, the foundation that is most famous for the annual cycling event on Mount Alpe d'Huez and that raised over 150 million euro for the fight against cancer. He works at the Central Bank of the Netherlands as a program manager and advisor for complex and politically difficult problems. His employer facilitates him in this job. His job enables him to be genuinely independent and to work tirelessly for the interests of all patients globally. He was honoured with a doctorate in October 2012 at the Free University in Amsterdam for connecting patients, researchers and clinicians all over the world.

Flash talks by young health innovators

Chair

Mauro Giacca

Professor of Cardiovascular Sciences, King's College London, United Kingdom

Mauro Giacca is a Professor of Cardiovascular Sciences at the School of Cardiovascular Medicine & Sciences, King's College London. Until 2018, he served as the Director-General of the International Centre for Genetic Engineering and Biotechnology (ICGEB), an international organization in the United Nations Common System. He is the President-Elect of the International Society for Heart Research (ISHR) - European Section and has served in the scientific councils of several biotechnology centres internationally. A medical doctor by training, he is a scientist active in the field of molecular medicine. His research interests focus on the development of novel biotherapeutics for degenerative disorders, in particular, myocardial infarction and heart failure.

Speaker

Dominik Schumacher

Group Leader and Founder, Tubulis Technologies, Germany

Dominik Schumacher studied business chemistry and chemistry at the University of Düsseldorf and conducted his PhD under the supervision of Christian Hackenberger at the Leibniz-Institut für Molekulare Pharmakologie and the Humboldt Universität zu Berlin. Together with his business partner, Jonas Helma-Smets, he is co-leading the award-winning start-up Tubulis, which has created a new technology-driven approach to accelerate and de-risk drug development with a specific focus on Antibody Drug Conjugates (ADCs) for targeted therapies. Dominik Schumacher filed several patents and is the first and corresponding author of publications in renowned journals including Angewandte Chemie, Chemical Science and Nature Chemistry. He received several prizes and was recently awarded as one of Europe's top innovators under 35.

Speaker

Francesco Gatto

Co-Founder, Chief Scientific Officer, Elypta, Sweden

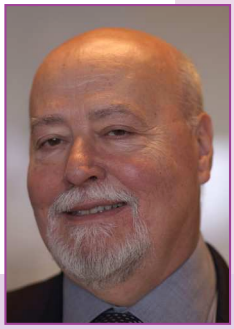
Francesco Gatto is the co-founder and Chief Scientific Officer at Elypta, a molecular diagnostics company based in Stockholm, Sweden. Francesco obtained an M.Sc. in Chemical Engineering from the University of Padova, Italy in 2011 and a PhD. in Systems Biology and Bioinformatics from Chalmers University of Technology, Sweden in 2015. He was a visiting scholar at University of California, San Diego in 2016. In 2017, he co-founded Elypta. Francesco Gatto is a co-inventor in the 3 patent applications that culminated in the creation of Elypta, whose core technology revolves around an innovative liquid biopsy for cancer, possibly the first one based on metabolism. For this venture, he attracted over €5M in research grants and private equity. He is the first author of 10 scientific publications since 2014. For his achievements with Elypta, Francesco was named MIT Technology Review Innovators Under 35 Europe in 2018, with a special mention as Pioneer of the Year 2018.

Session II:

Genomics - Opportunities and challenges

Chair

Giorgio Stanta
Professor of Pathology, University of Trieste, Italy



Giorgio Stanta is the head of the Molecular Histopathology Lab at the University of Trieste. He is the coordinator of the European group "Archive Tissues: Improving Molecular Medicine Research and Clinical Practice – IMPACTS" (www.impactsnetwork.eu). Also, he is the chairman of the "Biobanking and Molecular Pathobiology Working Group" of the OECI (Organisation of European Cancer Institutes – www.oeci.eu) and chairman of the "Molecular Pathology Working Group" of the ESP (European Society of Pathology – www.esp-pathology.org). He is a member of the managing board of BBMRI.IT (Italian Biobanking Infrastructure) and a member of the Committee of CEN (European Committee for Standardization) for molecular in-vitro diagnostic examinations.

Co-chair

Irene Norstedt
Acting Director, European Commission, Directorate-General for Research and Innovation, Belgium

Irene Norstedt is the Acting Director responsible for the Health Directorate of the DG for Research and Innovation at the European Commission. She is also Head of the Innovative and Personalised Medicine Unit. She has served at the European Commission since 1996 and was instrumental in the creation of the Innovative Medicines Initiative (IMI) in 2008. From 16 December 2014 to15 September 2015, Irene served as Acting Executive Director of the Innovative Medicines Initiative. Before joining the European Commission, she worked for the Swedish Life Science company, Biacore AB and at the Swedish Embassy in London. Irene studied biotechnology and polymer science, and holds a Master of Science (MSc) in Chemical Engineering.

Rapporteur

Rolf Apweiler

Director, The European Molecular Biology Laboratory - European Bioinformatics Institute, (EMBL-EBI), United Kingdom

Rolf Apweiler is Director of EMBL-EBI, together with Ewan Birney. Prior to this position, he was a Joint Associate Director, after many years of leading protein resources such as UniProt. Rolf Apweiler has made a major contribution to methods for the automatic annotation of proteins, making it possible to add relevant information to proteome sets for entire organisms. He spearheaded the development of standards for proteomics data, and his teams have maintained significant collections of protein identifications from proteomics experiments (PRIDE) and molecular interactions (IntAct). He also led EMBL-EBI's contribution to the Gene Ontology and is the current Director of Open Targets. Rolf received his PhD from the University of Heidelberg in 1994, and has been at EMBL since 1987. His major contribution to the field of proteomics was recognised by the Human Proteomics Organisation's "Distinguished Achievement Award in Proteomics" in 2004 and he was elected on as President of the Human Proteomics Organisation, which he held in 2007 and 2008. In 2012, he was elected as a member of EMBO and in 2015 he was elected to the ISCB (International Society for Computational Biology) as a fellow. Rolf Apweiler also served for many years on a multitude of Editorial Boards and Scientific Advisory Boards.

Keynote speaker

Jan Korbel

Group Leader, European Molecular Biology Laboratory (EMBL), Germany

Jan Korbel is a Group Leader and Senior Scientist at the EMBL. He holds a PhD in Molecular Biology, and during his postdoc at Yale University, he developed the paired-end mapping methodology for characterizing structural variations by next-generation sequencing. Jan Korbel has expertise in Human Genetics and Computational Biology and his research interests focus on understanding the determinants of genomic DNA rearrangement formation and selection in the germline, as well as in cancer genomes. He has had leading roles in the 1000 Genomes Project and the PCAWG Initiative. Also, he is significantly involved in a project dealing with scientific self-regulation in the context of whole-genome sequencing in patients and associated bioethical and normative aspects. Jan Korbel was elected into the German National Academy of Sciences Leopoldina in 2015 and into EMBO in 2016. He received the EACR – Pezcoller Foundation Cancer Research Award in 2018.

Keynote speaker

Barend Mons

Professor of BioSemantics, Leiden University Medical Center (LUMC), The Netherlands

Barend Mons is a global expert on FAIR principles. Originally a molecular biologist, he refocused in 2000 on semantic technologies and later on Open Science. In 2014, Barend Mons initiated the FAIR data initiative, and in 2015, he was appointed Chair of the European Commission's High-Level Expert Group for the "European Open Science Cloud", where he retired by the end of 2016. He continues to be active towards the practical realisation of the EOSC, defined as the Internet of FAIR data and services. Presently, Barend Mons is co-leading the GO FAIR initiative, an initiative to kick start developments towards the Internet of FAIR data and services, which will also contribute to the implementation of components of the European Open Science Cloud.

Panellist

Rodrigo Dienstmann

Principal Investigator, Vall d'Hebron Institute of Oncology (VHIO), Spain

Rodrigo Dienstmann is a medical oncologist with expertise in drug-biomarker co-development, computational research and real-world data analysis. He currently leads the Oncology Data Science Group of Vall d'Hebron Institute of Oncology, integrating clinical and translational research with genomics and immunophenotyping of tumors for precision cancer therapy. Rodrigo Dienstmann coordinates the Molecular Prescreening program of the institution, one of the largest in Europe, matching patients' tumor molecular alterations with targeted therapies and immunotherapies in early clinical trials. In parallel, he collaborates with Sage Bionetworks on biomedical research projects that bring cognitive computing closer to the clinics.

Panellist

Stefan Fröhling

Acting Managing Director, National Center for Tumor Diseases (NCT), Germany

Stefan Fröhling heads the Division of Translational Medical Oncology at DKFZ and is Acting Managing Director of NCT Heidelberg. Stefan Fröhling's research aims at providing patients with individually tailored cancer treatments. As part of the cross-institutional MASTER initiative of NCT Heidelberg/Dresden and the German Cancer Consortium, much of his work is centred on the development of tools for the comprehensive molecular and functional characterization of individual tumors, and the conception of clinical studies examining the efficacy of modern, molecularly targeted treatment approaches. In addition to his research in the precision oncology field, Stefan Fröhling particularly focuses on the biology of bone and soft-tissue sarcomas, as well as acute forms of leukaemia.

Panellist

Serena Scollen

Head of Department, European Life-Science Infrastructure For Biological Information (ELIXIR), United Kingdom

Serena Scollen is the Head of Human Genomics and Translational Data at ELIXIR, the European infrastructure for bioinformatics and life-science data, based in Hinxton, UK. Her vision is to ensure data that can be shared, will be shared responsibly. She works with scientists across Europe to establish standards and infrastructure to facilitate discoverability, access, sharing and analysis of genomics data, linked to other data types and at a scale that has not previously been achieved. Developing infrastructure will unleash new possibilities for genomics and health. Serena is a PI for the Innovative Medicine Initiative (IMI) FAIRplus project, an €8.3M collaboration that sets out to improve data sharing and reuse in life science research. Prior to joining ELIXIR, she was a Director within the Human Genetics and Computational Biomedicine group at Pfizer. In this role, she led and implemented a genetic and precision medicine strategy to support drug target selection and clinical programmes for the Pain and Sensory Disorders Research Unit. She was also a member of the ABPI Stratified Medicine Working Group. Earlier in her career, she worked within the Toxicogenomics group at GlaxoSmithKline. She gained postdoctoral experience at the University of Cambridge and Imperial College London and a PhD from the University of Cambridge, with a focus on the genetic susceptibility to disease.

Session III:

The public health perspective

Chair

Tit Albreht
Head of Centre, National Institute of Public Health (NIJZ), Slovenia

Tit Albreht is a Senior Health Services and Health Systems Researcher at the National Institute of Public Health of Slovenia and specialises in Social Medicine. He received his PhD from the University of Amsterdam, focussing his research on the exploration of the health services nationally and internationally. His research interests include the performance of health systems, evidence-based health policy, health workforce planning and mobility and development of more efficient health care, especially in the field of non-communicable diseases. He is an associate professor of public health in the Medical Faculty in Ljubljana and a member of the Scientific Committee of the European Public Health Association. He is a reviewer in several international and national medical and public health scientific journals. Since 2006 he has been actively involved in the development of cancer policies at the European level – during the Slovenian presidency to the Council of the EU and later as coordinator of Joint Actions on cancer policies at the EU level – European Partnership for Action Against Cancer (EPAAC), Cancer Control (CanCon) and the most recent one, innovative Partnership for Action Against Cancer (iPAAC). He worked in a number of projects for policy support in different countries in South-eastern Europe, e.g. Bosnia and Herzegovina, Macedonia, Moldova, Montenegro, Serbia and also in Kazakhstan. These consultancies included: support in the development of legal acts in the area of public health, cancer control and public health intervention. All of these aimed at supporting the policies of the respective ministries of health.

Co-chair

Elke Anklam
Director, European Commission, Directorate-General Joint Research Centre, Belgium

Elke Anklam is a chemist, with specialisation in food, organic and radiation chemistry. After obtaining her PhD from the University Hamburg (Germany), she worked in various European Research Institutions and was a Teaching Professor at the Applied University of Fulda (Germany). Since 1991, she has been working at the European Commission's Joint Research Centre (EC-JRC) where since 2006, she has been a Director at the JRC. Currently, she is the Director of the JRC-Geel site and Director of JRC Directorate F: Health, Consumers & Reference Material, located at the JRC-Geel and JRC-Ispra sites.

Rapporteur

Marc Van den Bulcke

Head of Service, Sciensano, Belgium

Marc Van den Bulcke obtained his PhD in sciences at the Laboratory of Genetics, University of Ghent (Belgium). He worked for about 7 years within the biotech industry at Plant Genetic Systems (now Bayer Crop Science) where he was involved in regulatory affairs and biotech product quality assurance. During this period, he gained real-life experience with global registration processes, accreditation, product quality control/assurance and business relationships. Since 2003, he works at the Scientific Institute of Public Health (Brussels, Belgium), now Sciensano, where he supported the establishment of the National Reference Laboratory on GMO detection. He worked for 3 years as a contract agent at the 'Institute of Health and Consumer Protection' at the Joint Research Centre of the European Commission where he coordinated policy-support research in molecular detection. Since 2013, he heads the Cancer Centre, a unit dedicated to the monitoring and evaluation of the activities on cancer control in Belgium. Current key activities of the Cancer Centre focus on the introduction of Next-Generation-Sequencing (NGS) in routine diagnostics in Belgium, on models to support the socio-professional reintegration of cancer patients, on screening and early detection (expert focus on cervical cancer) and on integrating patients, citizens and general practitioners views in the policy support activities of the Cancer Centre.

Keynote speaker

Thierry Philip

Head of the Institute Curie and President of Organisation of European Cancer Institutes (OECI), France

Thierry Philip is a Paediatrician and a specialist in bone marrow transplantation and immunology. Currently, he doubles as the Head of Curie Institute and President of the European Organisation of Cancer Institutes (OECI) and the Vice President of the Rhone Alpes Region and Lyon Metropole. Prior to this position, Thierry Philip was the Director of the Cancer Centre Lyon between 1989 and 2009.

Panellist

Nazneen Rahman

Independent healthcare consultant, United Kingdom

Nazneen Rahman's research integrates her medical and scientific expertise to identify and clinically implement human disease genes, with a major focus on cancer predisposition genes. She was Head of the Division of Genetics and Epidemiology at the Institute of Cancer Research, London and Head of Cancer Genetics at the Royal Marsden NHS Trust for 10 years, spanning to 2018. Nazneen Rahman was also Founder and Director of TGLclinical, a genetic testing lab using new sequencing and analytic technologies to deliver fast, affordable, cancer gene testing to the NHS. Nazneen Rahman has a strong commitment to open science and science communication and has garnered numerous awards, including a CBE. Nazneen Rahman is an AstraZeneca Non-Executive Director and chairs its Science Committee and is an independent healthcare consultant.

Panellist

Jacek Gronwald

Professor, Pomeranian Medical University (PMU), Poland

Jacek Gronwald is a professor at the Pomeranian Medical University, Poland and a specialist in gynaecology-obstetrics and clinical genetics. He is also a Regional Consultant in Clinical Genetics. From 2008 to 2016, he was the Dean of the Faculty of Medicine, Biotechnology and Laboratory Medicine at the Pomeranian Medical University (PMU).In 2008, he researched on Habilitation in the field of oncological genetics at the Pomeranian Medical University. Jacek Gronwald received his PhD in the field of oncological genetics from the Pomeranian Medical University, Poland in 1997. His research interests include cancer genetics; risk estimation, prevention and treatment of hereditary breast and ovarian cancer. Jacek Gronwald is an author and co-author of more than 250 papers mainly in the field of cancer genetics with IF of over 1500, and more than 9000 citations, including 43 h-index. He was born on 07 August 1965 in Koszalin, Poland.

Panellist

Jan-Ingvar Jönsson

Secretary General, Chair ICPerMed, Swedish Research Council, Sweden

Jan-Ingvar Jönsson obtained his PhD from Max-Planck-Institute in Freiburg, Germany, in 1990. After working as a postdoc at a Hospital for Sick Children in Toronto, he established a career as an independent investigator in experimental hematology and cancer biology in Sweden in the mid- 90's. Jan-Ingvar is currently the Secretary-General for Medicine and Health at the Swedish Research Council. His main responsibilities are the development of high-quality research programs and funding schemes in medicine, life science and clinical research. He is also involved in different aspects of assessment and impact of research within the health care sector. Since 2016, Jan-Ingvar is vice-chair of Joint Programming Initiative on Antimicrobial Resistance (JPIAMR). He was elected the new Chair of International Consortium of Personalised Medicine in November 2018. He is also chair of the joint committee of the Nordic medical research councils (NOS-M).

Panellist

Laura van't Veer

Director, University of California San Francisco (UCSF), Helen Diller Family Comprehensive Cancer Center, USA

Laura van 't Veer has a 25-year track record in molecular oncology research and over 280 scientific publications. She is Chair of the AACR Diagnostic Policy Committee and Chair of the Scientific Advisory Committee of the National Biomarker Development Alliance. She received the European Society Medical Oncology (ESMO) Lifetime Achievement Award for Translational Research in Breast Cancer in 2007, the 2014 European Union Prize for Women Innovators and the EPO European Inventor Award in 2015. In 2017, she received the European CanCer Organization (ECCO) Clinical Research Award. Last year, 2018, she was recognized by 24/7 Wall Street as one of '32 Amazing Women Inventors'. Laura van 't Veer holds a PhD in Medicine from the University of Leiden and has held several positions at the Cancer Center of Harvard Medical School, Massachusetts General Hospital in Boston and The Netherlands Cancer Institute where she headed the Diagnostic Oncology department. She is a Professor of Laboratory Medicine at UCSF since 2010. Laura van 't Veer is best known for her work that originated at the Netherlands Cancer Institute (NKI) on a 70-gene activity signature which distinguishes whether breast cancer has a low or high risk of recurrence. This was the basis for the MammaPrint® test (Agendia, co-founded by Dr van't Veer). MammaPrint® obtained clearance from the FDA in 2007 and is included in many national and international guidelines.

Flash talks:

A focus on genomics for rare diseases

Chair

Julia Wilson
Associate Director, Wellcome Trust Sanger Institute, United Kingdom

Julia builds and supports the relationships to support the strategic vision of the Sanger Institute. These activities range from interactions with the academic scientific community, commercial partners, funders and other key stakeholders. She increases awareness of the Sanger Institute's research, explores new collaborations and areas of research. She works in the position of serving as a source of translational opportunities, raises awareness of the importance and impact of genomics research with government and policymakers, and facilitates the translation of genomics research into clinical practice at the Sanger Institute. Previously, Julia was Assistant Director of Research at Breakthrough Breast Cancer and Science Programme Manager at the World Cancer Research Fund. As a scientist, she was a post-doc at the Karolinska Institute in Sweden and then worked as a cancer researcher at Cancer Research UK and the Queen Mary University of London.

Speaker

Han Brunner
Head of Department, Radboud University Medical Center (Radboudumc), The Netherlands

Han Brunner is a full professor and Head of the Department of Human Genetics at Nijmegen University Hospital, and Maastricht University Medical Center, in the Netherlands. He served as board member of the Dutch, European (president in 2014-2015) and American Societies of Human Genetics. He was elected member of the Royal Netherlands Academy of Arts and Sciences, and the Academia Europea. He is a Knight in the Order of the Dutch Lion since 2013. He received the King Faisal International Prize in Medicine 2016, the 2017 Carter medal of the British Clinical Genetics Society, and the Medal of Honour of the German Society of Human Genetics. Han Brunner discovered a large number of disease genes, and by applying cutting- technologies (genomic microarrays, exome sequencing, and whole genome sequencing) to understand genetic diseases. Much of his work is on neurodevelopmental conditions such as intellectual disability and abnormal behaviour. His work has established that in non-consanguineous populations, the major cause of intellectual disability lies in spontaneous new mutations.

Speaker

Daria Julkowska
Assistant Director, National Institute of Health and Medical Research (INSERM), France

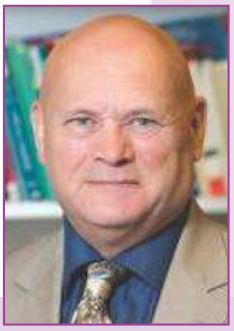
Daria Julkowska is the Assistant Director of the Institute of Genetics, Genomics and Bioinformatics at INSERM. She is also the coordinator of the European Joint Programme Cofund on Rare Diseases (EJP RD) that brings together 130 institutions from 35 European, Associated countries and Canada. Previously, she was the Scientific Coordinator at ANR responsible for the management of several EU and international funding programmes, including the ERA-Net E-Rare. She developed and implemented a set of collaborations facilitating rare diseases research, including partnerships with EU Research Infrastructures and Patients' Organizations. She has extensive knowledge of the European funding schemes and programmes. Since February 2017, she has been serving as Chair of the Funders Constituent Committee of IRDiRC.

Session IV:

The citizen and patient perspective

Chair

Peter Goodhand
Chief Executive Officer, Global Alliance for Genomics and Health, Canada



Peter Goodhand is a leader in the global health sector as a senior executive and board member. He played a key role in the creation of the Global Alliance for Genomics and Health (GA4GH) and was appointed as its founding Executive Director in 2014, and as Chief Executive Officer in 2018. From May 2016 to April 2018, he also served as the President of the Ontario Institute for Cancer Research (OICR). Prior to the GA4GH and OICR, he was the President and Chief Executive Officer of the Canadian Cancer Society, Canada's largest health charity. Before joining the charitable sector, Goodhand had a 20 year career in the global medical technology industry, including strategic leadership roles with multinational healthcare companies such as American Cyanamid and Johnson & Johnson; Board Chair and President of Canada's Medical Device Industry association (MEDEC); and as the founding Managing Director and then Board Chair of the Health Technology Exchange (HTX). Peter Goodhand is currently a member of the Occupational Cancer Research Centre Steering Committee, Co-chair of the Medical and Scientific Advisory Board of Global Genes, Co-chair of the International 100K+ Cohorts Consortium (IHCC), and a member of the of the Global Genomic Medicine Collaboration (G2MC) Steering Committee. He chaired the Government of Canada's Expert working group on the future of medical isotope production, and was a member of the Canadian delegation to the UN summit on non-communicable diseases.

Co-chair

Ioana-Maria Gligor
Head of Unit, European Commission, Directorate-General for Health and Food Safety, Belgium

Ioana-Maria Gligor is Head of Unit of the European Reference Networks and Digital Health at the European Commission, in DG Health and Food Safety (SANTE). Before joining SANTE, Ioana was Deputy Head of Unit in the Secretariat General of the Commission and at DG Employment and Social Affairs (EMPL) and assistant to the Director General of DG EMPL. She began her career at the European Commission in the cabinet of Commissioner Orban. Before joining the European administration, she was the spokesperson for the Romanian Chief Negotiator with the EU. Her background is in EU affairs and political sciences, combined with an IT- high school acquired expertise.

Rapporteur

Rita Schmutzler
Head of Department, University Hospital Cologne, Germany

Rita Schmutzler is Director of the Center for Familial Breast and Ovarian Cancer, University Hospital Cologne and coordinator of the German Consortium for Hereditary Breast and Ovarian Cancer. She did her specialist training in Obstetrics and Gynecology and received her postdoctoral lecturing qualification (Habilitation) from the University of Bonn in 1998 and the Federal Licensing Examination (FLEX) USA in 1997. She is a member of numerous committees such as the expert panel of the National Cancer Plan at the Federal Ministry of Health, the S3 guideline committee on breast cancer and the committee of the gene diagnostic act at the Robert-Koch Institute, as well as a member of some scientific advisory boards e.g. at the Federal Institute for Drugs and Medical Devices (BfArM) and the Institute for Quality and Efficiency in Health Care (IQWIG). Her main research focus is to understand the hereditary basis of breast and ovarian cancer, identify genotype-/phenotype correlations and translate these findings into risk-adjusted clinical prevention programs.

Keynote speaker

Bartha Knoppers

Director, McGill University, Canada

Bartha Maria Knoppers, PhD (Comparative Medical Law), is a Full Professor, Canada Research Chair in Law and Medicine and Director of the Centre of Genomics and Policy of the Faculty of Medicine at McGill University. She is Chair of the Ethics and Governance Committee of the International Cancer Genome Consortium (2009-2017), as well as the Ethics Advisory Panel of WADA (2015-). She is Co-Chair of the Regulatory and Ethics Workstream of the Global Alliance for Genomics and Health (2013-). From 2015-2016, she was a member of the Drafting Group for the Recommendation of the OECD Council on Health Data Governance and gave The Galton Lecture in November 2017. She holds four Doctorates Honoris Causa and is a Fellow of the American Association for the Advancement of Science (AAAS), the Hastings Center (bioethics), the Canadian Academy Health Sciences (CAHS), and, the Royal Society of Canada. She is also an Officer of the Order of Canada and of Quebec.

Keynote speaker

Francesco Florindi

Engagement Officer, Biobanking and Biomolecular Resources Research Infrastructure - European Research Infrastructure Consortium (BBMRI-ERIC), Austria

Francesco Florindi obtained a cum laude Master's degree in International Relations and Diplomacy from the University of Trieste-Gorizia with a thesis on EU enlargement. In 2011, he started working for regional representatives, NGOs, and the European Commission's Joint Research Centre (JRC) in Brussels. He went on to join the European Cancer Patient Coalition as Public Affairs Coordinator and became Head of EU Affairs in 2016. His experience working in healthcare dates back to 2013 when he joined the ECCO and SIOPE Public Affairs team. Francesco has worked on a number of key European issues such as data protection, health technology assessment, access to quality healthcare, eHealth/mHealth and patient advocacy. These experiences made him understand how patients and healthcare professionals can fruitfully collaborate at European and international level in order to reach common goals. Francesco Florindi is a fellow Young Gasteiner and a Member of the European Health Parliament. He is an Italian and speaks English and French.

Panellist

Jane Kaye

Director, Centre for Law, Health and Emerging Technologies (HeLEX), University of Oxford, United Kingdom

Jane Kaye is the Director of the Centre for Law, Health and Emerging Technologies (HeLEX) at the University of Oxford. She is also a part-time Professor at the University of Melbourne, Australia where she has a research team. She obtained her degrees from the Australian National University (BA); University of Melbourne (LLB); and University of Oxford (DPhil). She serves on several international expert committees and scientific advisory boards and is on the editorial boards of Law, Innovation and Technology, of the Journal of Law, Information and Science, of New Genetics & Society and of Life Sciences, Society and Policy. Her team are leading on the Dynamic Consent project, and she is one of the leaders in the ELSI 2.0 Global Initiative. Her research focuses on the relationships between law, ethics and the emerging technologies in health. The main focus of her research is on genomics with an emphasis on biobanks, privacy, data-sharing frameworks, global governance and translational research.

Panellist

Bettina Borisch

Professor of Social and Preventive Medicine, University of Geneva, Switzerland

Bettina Borisch is an MD and a Histopathologist, MPH and Fellow of the Royal College of Pathology (UK). Her scientific research work delves into neoplastic lesions of the immune system and breast cancer. Her interests also include community-based oncology, as well as health communication and global health. She is the Editor in Chief of "Pathobiology" and the Co-Editor of "Journal of Public Health Policy". In addition to her academic work, she serves as the Director of the World Federation of Public Health Associations and holds positions at several Committees of Public Health-oriented institutions. She was president of Europa Donna – The European Breast Cancer Forum – and Founding President of the Swiss Forum of Europa Donna. She teaches at the University of Geneva, the Swiss School of Public Health and she also teaches patient support groups. She is an author of more than 120 scientific papers and 2 books.

Panellist

Jean-Pierre Hubaux

Professor, Ecole Polytechnique Fédérale de Lausanne (EPFL), Switzerland

Jean-Pierre Hubaux is a Full Professor at Ecole Polytechnique Fédérale de Lausanne (EPFL). Through his research, he contributes to laying the foundations and developing the tools for protecting privacy in tomorrow's hyper-connected world. He has pioneered the areas of privacy and security in mobile/wireless networks and in genomics. He is the academic director of the recently created Center for Digital Trust (C4DT). He leads the ETH-funded project, Data Protection in Personalized Health (DPPH) and is a co-chair of the Data Security Work Stream of the Global Alliance for Genomics and Health (GA4GH). He is one of the seven commissioners of the Swiss FCC and is a Fellow at both IEEE (2008) and ACM (2010). He is among the most cited researchers in privacy protection and information security.

Panellist

Andres Metspalu

Head of Department, University of Tartu, Institute of Genomics, Estonia

Andres Metspalu obtained his MD in 1976, PhD in 1979 and did his postdoc at Colombia University and Yale University from 1981-1982. His main scientific interests are human genetics, genomics of complex diseases and population-based biobanks and application of the precision medicine in health care. From 1993-1994, he served as a visiting faculty staff at Baylor College of Medicine, Houston, and in 2000 he was a recipient of the International Visiting Senior Scientist Award offered by IARC, Lyon, France. From 1996 to 2008, Andres. Metspalu was also the Head and founder of the Molecular Diagnostic Center of the Tartu University Hospital. Andres Metspalu was previously the president of the ESHG in 2006. In 2010, he was elected to the Estonian Academy of Sciences. He serves in several national and international committees and has received among other awards and honours the Order of the Estonian Red Cross 3rd Class and L'Ordre des Palmes Académiques from the Republic of France. From 2010 he was awarded Doctor Honoris Causa from Vilnius University.

Final keynote presentation

Chair

Guy Van den Eede

Head of Unit, European Commission, Directorate-General Joint Research Centre, Belgium

Guy Van den Eede is an agricultural engineer, specialized in plant molecular biology. He has been appointed at the Joint Research Centre of the European Commission in 1990 to provide technical support to the implementation of the EU policies on GMOs; later he has set up and managed the European Union Reference Laboratory for GMOs. In 2016, he was appointed Unit Head Knowledge for Health and Consumer Safety, covering life science-related files in the field of e.g. public health, food safety and security, toxicology, molecular biology and GMOs. Attention is given to anticipating knowledge needs and mapping knowledge gaps. The Unit has staff on both the Belgian site in Geel and the Italian site in Ispra.

Keynote speaker

Teri Manolio

Director, National Human Genome Research Institute, National Institutes of Health (NIH), USA

Teri Manolio directs the NHGRI's Division of Genomic Medicine, where she leads programs to develop and implement genomic applications in clinical care. She came to NHGRI in 2005 to lead efforts in applying genomic technologies to population research, including the Electronic Medical Records and Genetics (eMERGE) Network, the NHGRI Genome-Wide Association Catalog, and the Clinical Genome (ClinGen) Resource. She continues to practice and teach internal medicine at the Walter Reed National Military Medical Center and the Uniformed Services University of the Health Sciences. She is the author of over 280 research publications and has research interests in genome-wide association studies of complex diseases, ethnic differences in disease risk, and incorporating genomic findings into clinical care.

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