
Monica Lanzoni, Agnieszka Kinsner-Ovaskainen (JRC), Joan Morris (Population Health Research Institute, St George’s, University of London, UK), Simona Martin (JRC)
Down syndrome accounts for 8% of all congenital anomalies. It is related to maternal age, which generally increased in Europe during the study period, with large differences in European regions. The total prevalence of Down syndrome for 10,000 births increased from 16 in 1990 to 23 in 2015. The prenatal detection increased from 49% in 2005 to about 70% in 2015, but territorial differences exist.

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Contact information
Monica Lanzoni, Agnieszka Kinsner-Ovaskainen
Directorate for Health, Consumers and Reference Materials
Health in Society Unit
Address: via Fermi, 2749 – Ispra (VA) – Italy
Email: monica.lanzoni@ec.europa.eu, agnieszka.kinsner-ovaskainen@ec.europa.eu

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The current political and economic challenges faced by the European Union and its Member States call even more for evidence-informed policies. They also require tailor-made policies, developed using highly sophisticated analyses based not only on country-level data, but rather on regional and sub-regional knowledge.

National averages, in particular, bear the risk to present a misleading picture in countries with significant disparities between different regions and areas.

Looking only at national averages can also limit and delay understanding of the differences between regions and cities – identifying leaders and laggards –, as well as prevent the identification of emerging trends in certain socio-economic indicators. Only a detailed analysis of data at regional and local level can bring these insights.

The Joint Research Centre (JRC) of the European Commission has developed the *Socio-economic regional microscope*. It is a new series of short periodical publications which aims to open-up new areas of analysis, and present the stories which can only be told using regional socio-economic data.

Each report presents EU socio-economic indicators according to a data storytelling principle, using a combination of three key elements: data, visuals (maps), and narrative. Each indicator will therefore be represented through maps at regional level (NUTS2), and in some cases even at the NUTS3 and local level.

The *Socio-economic regional microscope* will also show the breadth of the JRC regional analysis in a wide range of research areas: culture, economics, education, energy, healthcare, research and innovation, tourism, etc.

The reports, data and maps are also available on the Territorial Dashboard website of the JRC Knowledge Centre for Territorial Policies, in the *Thematic Analyses* section: [https://urban.jrc.ec.europa.eu/t-board/#/thematic-analyses](https://urban.jrc.ec.europa.eu/t-board/#/thematic-analyses).
Introduction

Structural defects (congenital malformations, deformations, disruptions and dysplasias) and chromosomal abnormalities are a major cause of infant mortality, childhood morbidity, long-term disability and among the leading causes of years of potential life lost. Congenital Anomalies carry a high burden to affected individuals, their families and the community in terms of quality of life, participation in the community and need for services.

EUROCAT\textsuperscript{1}, founded in 1979 as the European Concerted Action on Congenital Anomalies and Twins, is a high quality network of population-based registries\textsuperscript{2} across Europe for the monitoring, surveillance and research of congenital anomalies.

Currently, the JRC-EUROCAT Central Registry holds details on approximately 800 000 cases of congenital anomalies collected since 1980 by 51 member registries from 23 countries, as anonymous individual case specific data (full members) or aggregate prevalence data in tables (associate members). More than 1.4 million births are surveyed per year in Europe, in which is equivalent to 26 % of the EU-28 birth population being covered. EUROCAT produces statistics on outcomes and prevalence rates for a wide range of major congenital anomalies annually. The data cover all pregnancy outcomes including live births, stillbirths, late foetal deaths from 20 weeks of gestation and terminations of pregnancy for foetal anomalies (TOPFA).

\textsuperscript{1} From 2015, after the transfer of the Central Registry and European level coordinating activities to the EC’s Joint Research Centre, EUROCAT is an integral part of the European Platform on Rare Diseases Registration being developed by the JRC in close collaboration with the Directorate General for Health and Food Safety (DG SANTE).

\textsuperscript{2} Population-based type I registries: collect the outcome of births from mothers resident in the area covered by the registry, wherever the outcome occurred.
Down syndrome, a disorder caused by trisomy of chromosome 21, accounts for approximately 8% of all congenital anomalies. The total prevalence of Down syndrome (including all pregnancy outcomes) in the EUROCAT registries in the years 1990-2014 is presented in Figure 1 in the next page.

Down syndrome is associated with many physical anomalies and intellectual disabilities [1]. Over 43% of babies with Down syndrome have a major cardiac anomaly. Other major non-cardiac congenital anomalies are also frequent (15%), particularly in the digestive system and limbs (Table 1).

### Occurrence of Down syndrome and associated anomalies

<table>
<thead>
<tr>
<th>System</th>
<th>% of live birth and fetal death cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital Heart Defects</td>
<td>43.6%</td>
</tr>
<tr>
<td>Digestive system</td>
<td>7.0%</td>
</tr>
<tr>
<td>Limbs</td>
<td>3.3%</td>
</tr>
<tr>
<td>Urinary</td>
<td>1.9%</td>
</tr>
<tr>
<td>Nervous system</td>
<td>0.9%</td>
</tr>
<tr>
<td>Eye</td>
<td>1.6%</td>
</tr>
<tr>
<td>Genital</td>
<td>0.5%</td>
</tr>
<tr>
<td>Ear, face and neck</td>
<td>1.2%</td>
</tr>
<tr>
<td>Oro-facial clefts</td>
<td>0.4%</td>
</tr>
<tr>
<td>Respiratory</td>
<td>1.2%</td>
</tr>
<tr>
<td>Abdominal wall defects</td>
<td>0.3%</td>
</tr>
</tbody>
</table>

Table 1. Percentage of cases of Down syndrome (live birth and fetal death from 20 weeks of gestation) with at least one anomaly referred to a given system, which was reported by selected EUROCAT registries [1].

Note: The areas in white are not covered by the EUROCAT registries.
Down syndrome is known to be more prevalent in children born to older mothers. The overall increasing prevalence of Down syndrome during the last decades is mainly due to the increase in mean maternal age, because the age-specific prevalence remains constant with the distribution shown in Figure 2.

The average age of women giving birth has steadily increased in the last decades [2]. This tendency is evident also in the areas covered by EUROCAT.

Figure 3 shows that there is an increase in the percentage of mothers aged ≥35 in the population of the EUROCAT registries, but there is also a large variation in the maternal age distribution in European regions.

Note: The areas in white are not covered by the EUROCAT registries.
Differences in pregnancy outcomes and prenatal diagnosis of Down syndrome in Europe

An important public health indicator developed by EUROCAT [3] is the prevalence of Down syndrome in live born children, which has an impact on health service requirements and socio-economic implications. This indicator gives the combined effect of delayed childbearing, and of policies on prenatal screening and termination of pregnancy.

Although the total prevalence increased in the last two decades, the prevalence of live births overall remains stable, while there is a rise in the prevalence of TOPFA for Down syndrome (Figure 4).

There are significant regional differences in both total and live birth prevalence of Down syndrome. The variation between the different countries depends on the national policies regarding terminations of pregnancy for foetal anomalies, as well as on the availability and provision of prenatal screening.

In most countries where TOPFA is allowed, an increase in terminations for Down syndrome is observed in the last years (Figure 5).

Note: Cork and Kerry (Ireland) reported some TOPFA cases where termination was performed abroad. The areas in white are not covered by the EUROCAT registries.
National prenatal screening programs for Down syndrome have been established in many European countries. The possibilities of prenatal diagnosis of Down syndrome have changed in the last two decades, with the availability of new and more reliable methods (including non-invasive techniques, such as serum screening test or the cell-free foetal DNA detection in maternal blood) [4].

The increase in prenatal diagnosis at younger gestational ages contributes, in addition to the maternal age, to the increase of the overall prevalence identifying affected foetuses that, if not detected prenatally, would have resulted in an undiagnosed foetal loss at a later gestation.

The use of the different techniques/methods varies depending on the countries’ screening policy, screening related costs and reimbursement possibilities, as well as on the local policy on TOPFA (e.g. the use of some screening methods is not necessary and often not done if TOPFA is not allowed).

Figure 6 shows the distribution of the different screening techniques used for prenatal diagnosis of Down syndrome in the last ten years. There has been also an overall increase in the percentage of cases prenatally diagnosed in the last ten years.
**Acknowledgements**

We would like to acknowledge all the active and past EUROCAT registries ([http://www.eurocat-network.eu/pagecontent.aspx?tree=allmembers](http://www.eurocat-network.eu/pagecontent.aspx?tree=allmembers)) for providing the data for this report.

**Active EUROCAT registries:** Austria (Styria); Belgium (Antwerp, Hainaut-Namur); Croatia (Zagreb); Czech Republic; Denmark (Odense); Finland; France (Auvergne, Brittany, French West Indies, Isle de Réunion, Paris, Rhône-Alpes); Germany (Mainz, Saxony Anhalt); Hungary; Italy (Emilia Romagna, Tuscany); Ireland (Cork & Kerry, Dublin, South East Ireland); Malta; Netherlands (North Netherlands); Norway; Poland (Polish National Registry, Wielkopolska); Portugal (South Portugal); Spain (Basque Country, Valencia Region); Sweden; Switzerland (Vaud); Ukraine (OMNI-Net); United Kingdom (East Midlands & South Yorkshire, Northern England, South West England, Thames Valley, Wessex, Wales).

**Past EUROCAT registries:** Bulgaria (Sofia); France (Central East France, Strasbourg); Ireland (Galway); Italy (Campania, North East Italy, Sicily); Spain (Asturias, Barcelona); United Kingdom (Glasgow, North West Thames).

We thank also the **JRC-EUROCAT Management Committee** for critically reviewing this contribution.

**Acronyms**

- TOPFA - termination of pregnancy for foetal anomalies (Figure 5, explained in the text on page 4)
- GA – gestational age (Figure 6)
- CVS – chorionic villus sampling (Figure 6)

**References**


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