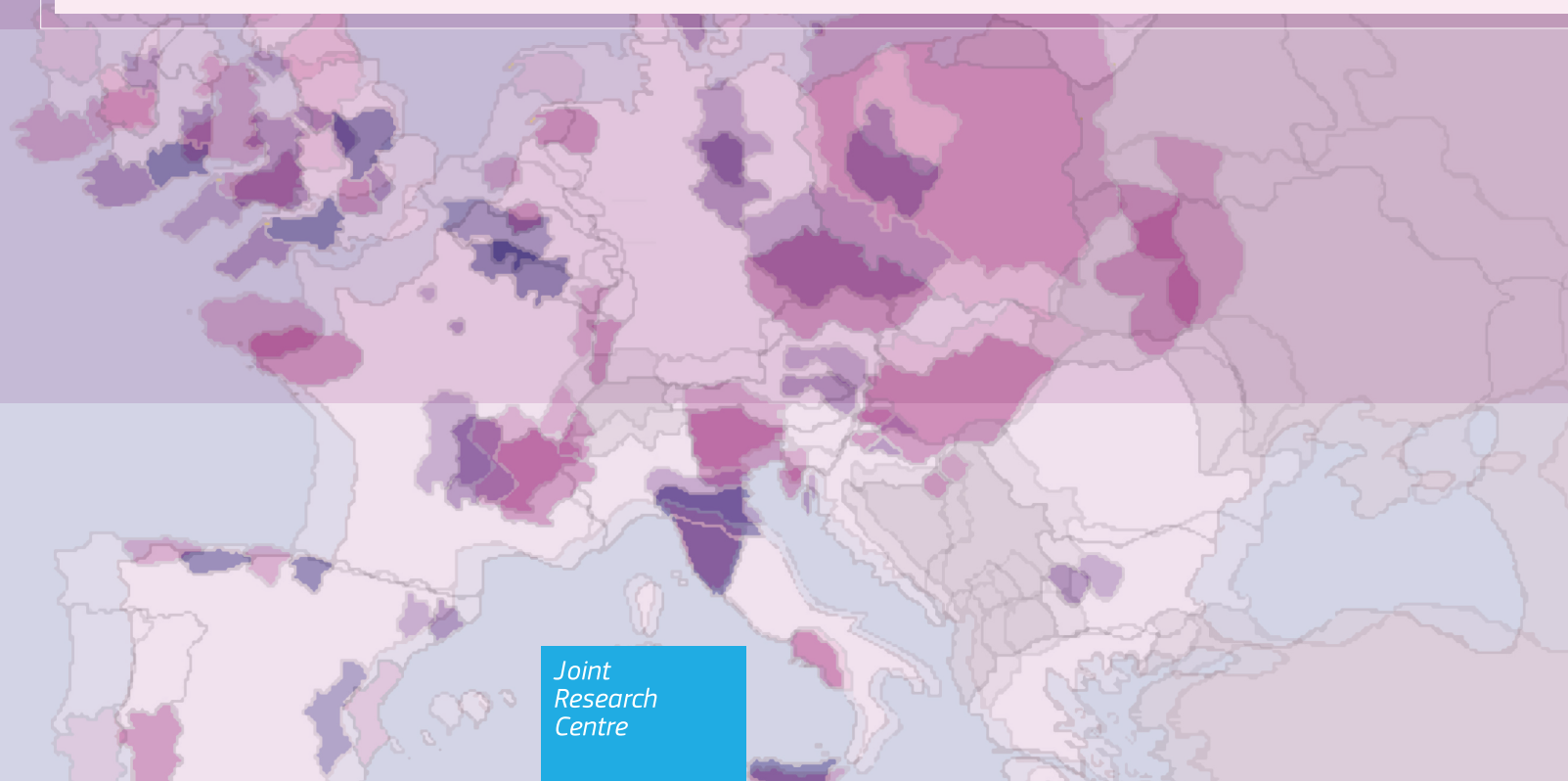
European Commission

Socio-economic regional microscope series

EUROCAT – Surveillance of congenital anomalies in Europe: epidemiology of Down syndrome 1990-2014

Monica Lanzoni, Agnieszka Kinsner-Ovaskainen (JRC), Joan Morris (Population Health Research Institute, St George's, University of London, UK), Simona Martin (JRC)



Joint Research Centre

European Commission
Joint Research Centre

Socio-economic regional microscope series

EUROCAT – Surveillance of congenital anomalies in Europe: epidemiology of Down syndrome 1990-2014

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.

Manuscript completed in February 2019

Luxembourg: Publications Office of the European Union, 2019

JRC115824

PDF ISBN 978-92-76-00574-2 ISSN 2599-6304 doi:10.2760/70796 KJ-BF-19-001-EN-N
Print ISBN 978-92-76-00575-9 ISSN 2599-6290 doi:10.2760/331810 KJ-BF-19-001-EN-C

How to cite this report: M. Lanzoni, A. Kinsner-Ovaskainen, J. Morris, S. Martin, *Socio-economic regional microscope series - EUROCAT – Surveillance of congenital anomalies in Europe: epidemiology of Down syndrome 1990-2014*, Publications Office of the European Union, Luxembourg, 2019, ISBN 978-92-76-00574-2, doi:10.2760/70796, JRC115824

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*

EU Science Hub
https://ec.europa.eu/jrc

Table of contents

The Socio-economic regional microscope series

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*.

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[1], founded in 1979 as the European Concerted Action on Congenital Anomalies and Twins, is a high quality network of population-based registries[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).

[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).

[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.

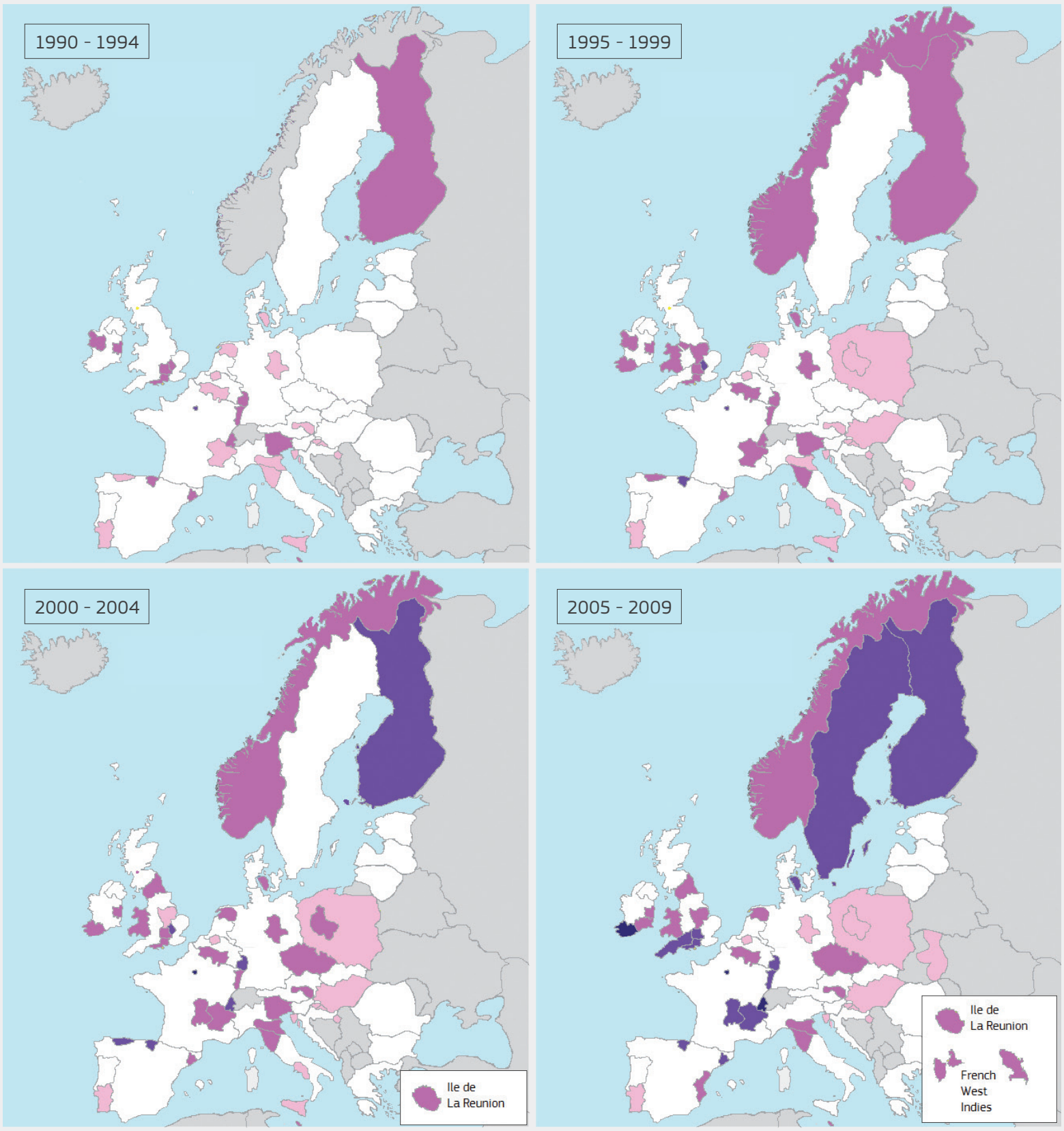
Occurrence of Down syndrome and associated anomalies

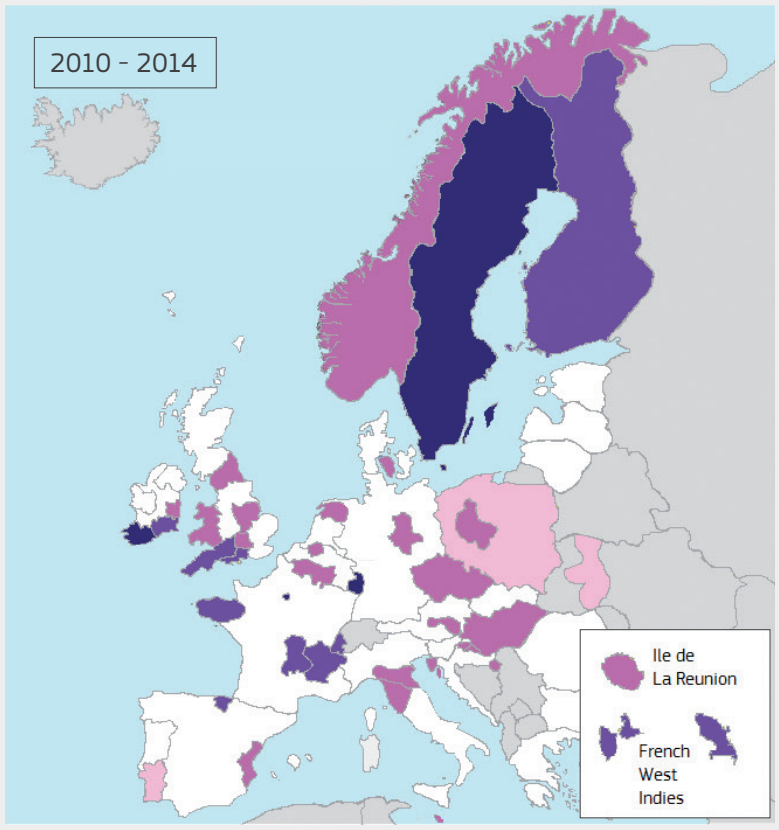
Down syndrome, a disorder caused by trisomy of chromosome 21, accounts for approximately 8 % of all congenital anomalies. The total prevalence of Down syndrome (including all pregnancy outcomes) in the EUROCAT registries in the years 1990-2014 is presented in Figure 1 in the next page.

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).

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

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].





Prevalence per 10 000 total births

- 0 - 15.9
- 16 - 25.9
- 26 - 35.9
- ≥ 36

Figure 1. Total prevalence of Down syndrome per 10.000 total births in the EUROCAT registries for five-years intervals (1990-1994, 1995-1999, 2000-2004, 2005-2009 and 2010-2014).

Note: The areas in white are not covered by the EUROCAT registries.

Prevalence of Down syndrome and increase in maternal age

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.

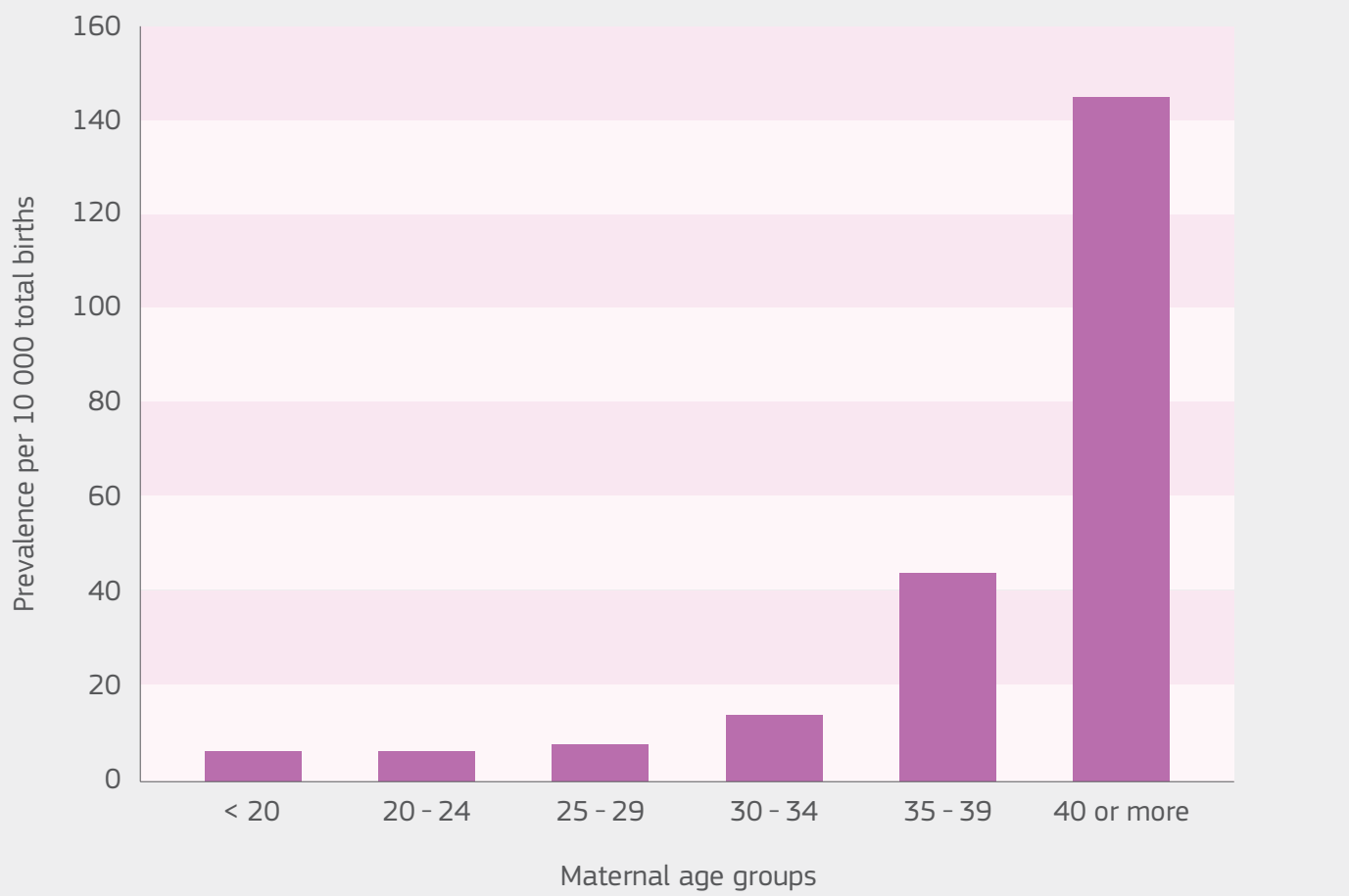
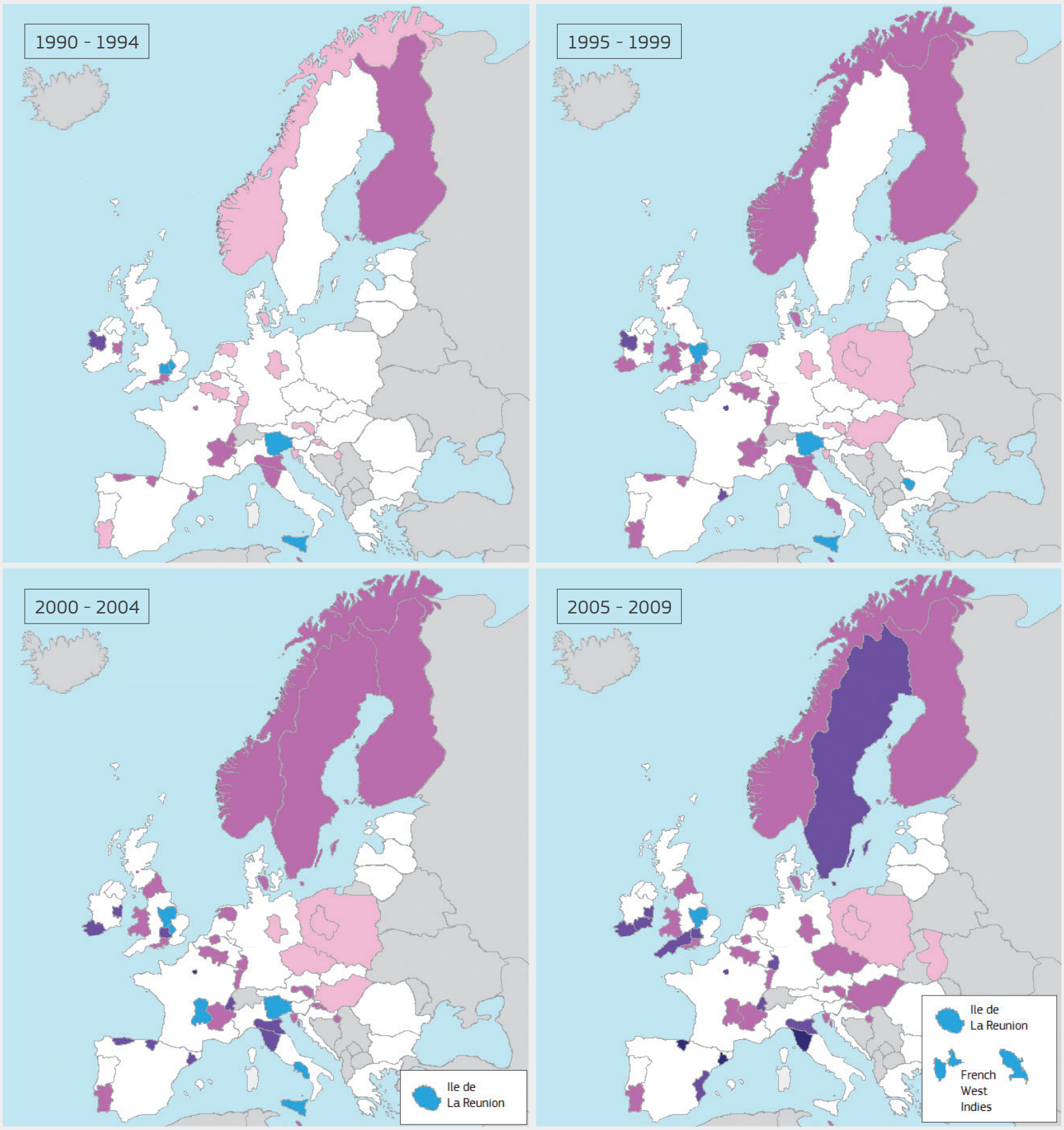


Figure 2. Total prevalence of Down syndrome in 10.000 total births by six maternal age groups (based on EUROCAT data 1990-2014).

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.



Percentage of mothers with age ≥35 in the population

- ≤ 10.9 %
- 11 - 20.9 %
- 21 - 30.9 %
- ≥ 31 %
- Data not availabale

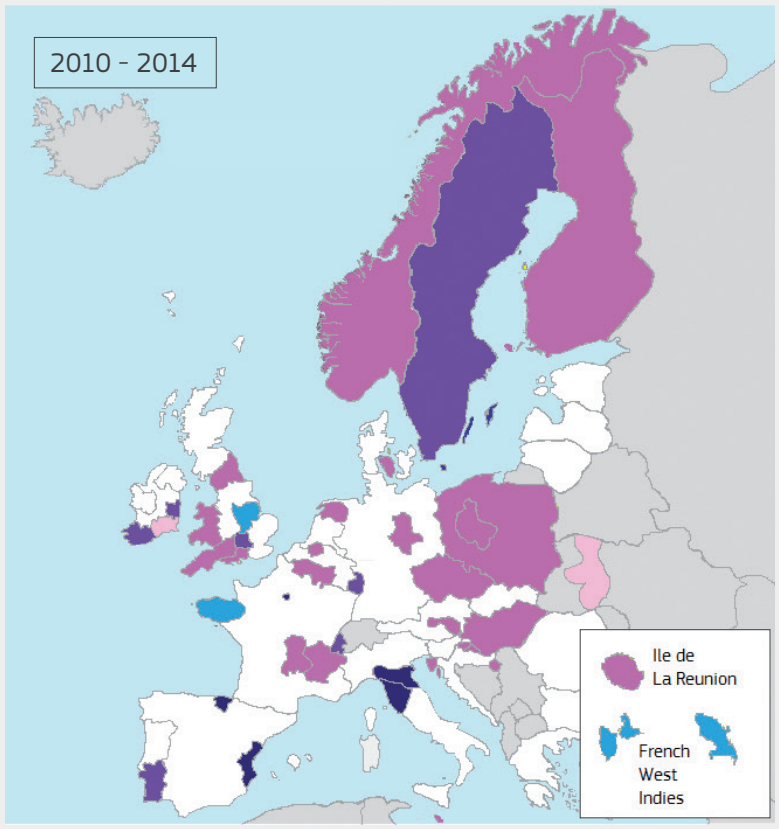


Figure 3. Percentage of mothers with age ≥35 in the population of the EUROCAT registries for five-years intervals (1990-1994, 1995-1999, 2000-2004, 2005-2009 and 2010-2014).

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).

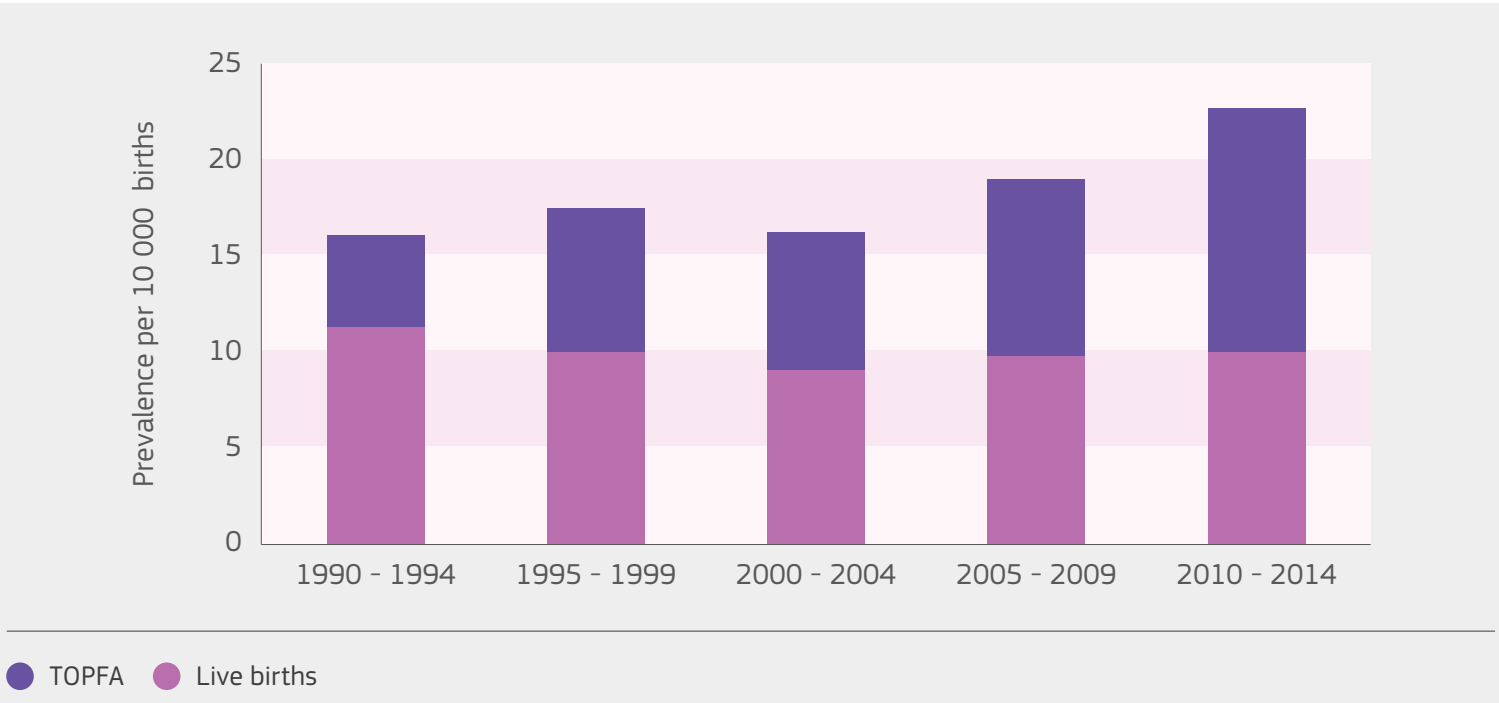
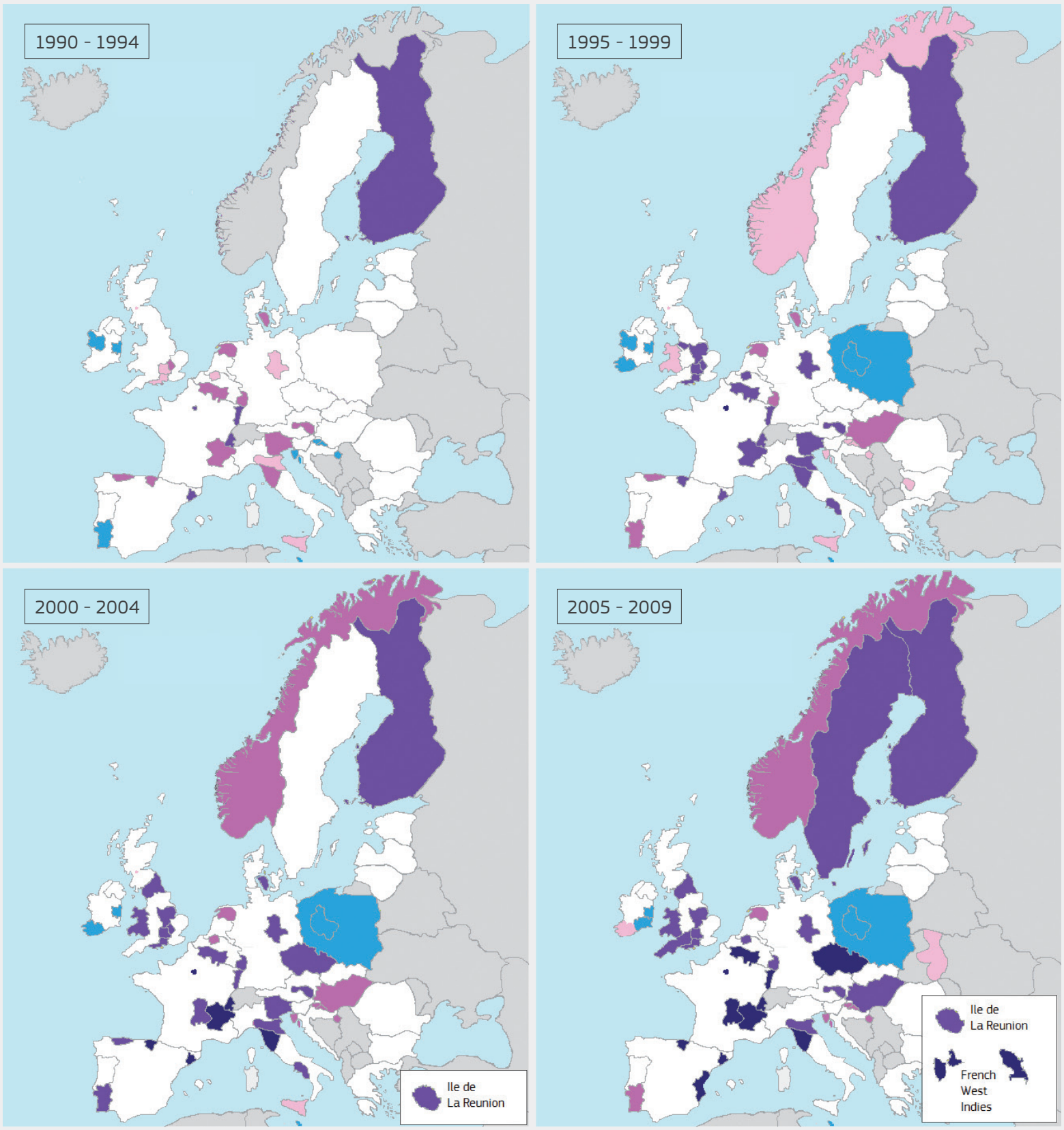


Figure 4. Total prevalence of Down syndrome in Europe per 10 000 total births in EUROCAT registries for 5-years intervals (1990-1994, 1995-1999, 2000-2004, 2005-2009, 2010-2014), according to outcome (live birth or TOPFA).

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).



Percentage of Down Syndrom diagnoses resulting in TOPFA

- ≤ 15.9 %
- 16 - 40.9 %
- 41 - 69.9 %
- ≥ 70 %
- TOPFA not allowed

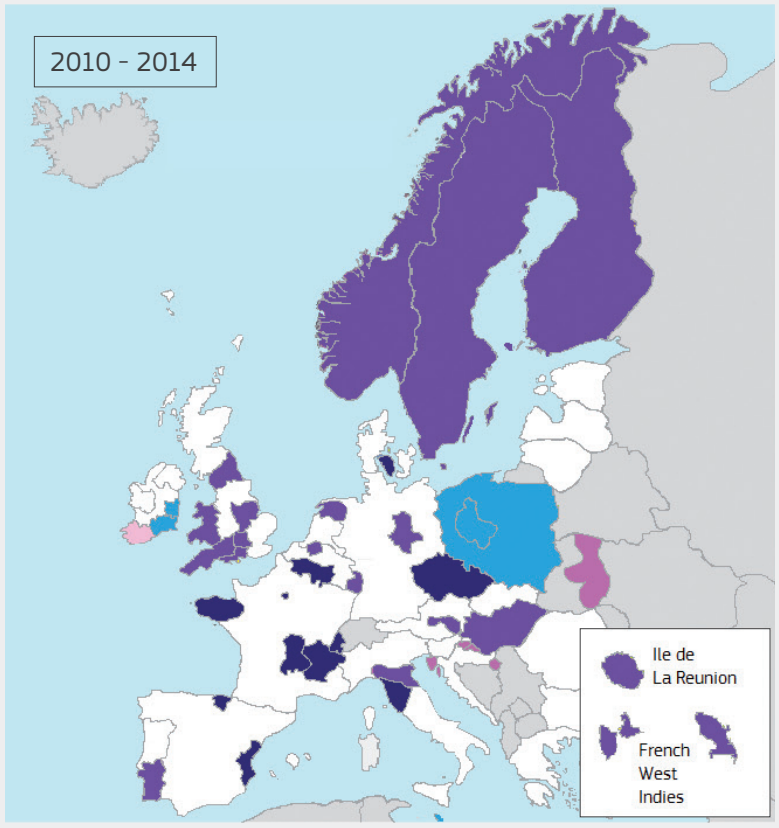


Figure 5. Percentage of Down syndrome diagnoses resulting in a TOPFA reported by the EUROCAT registries for five-years intervals (1990-1994, 1995-1999, 2000-2004, 2005-2009 and 2010-2014).

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.

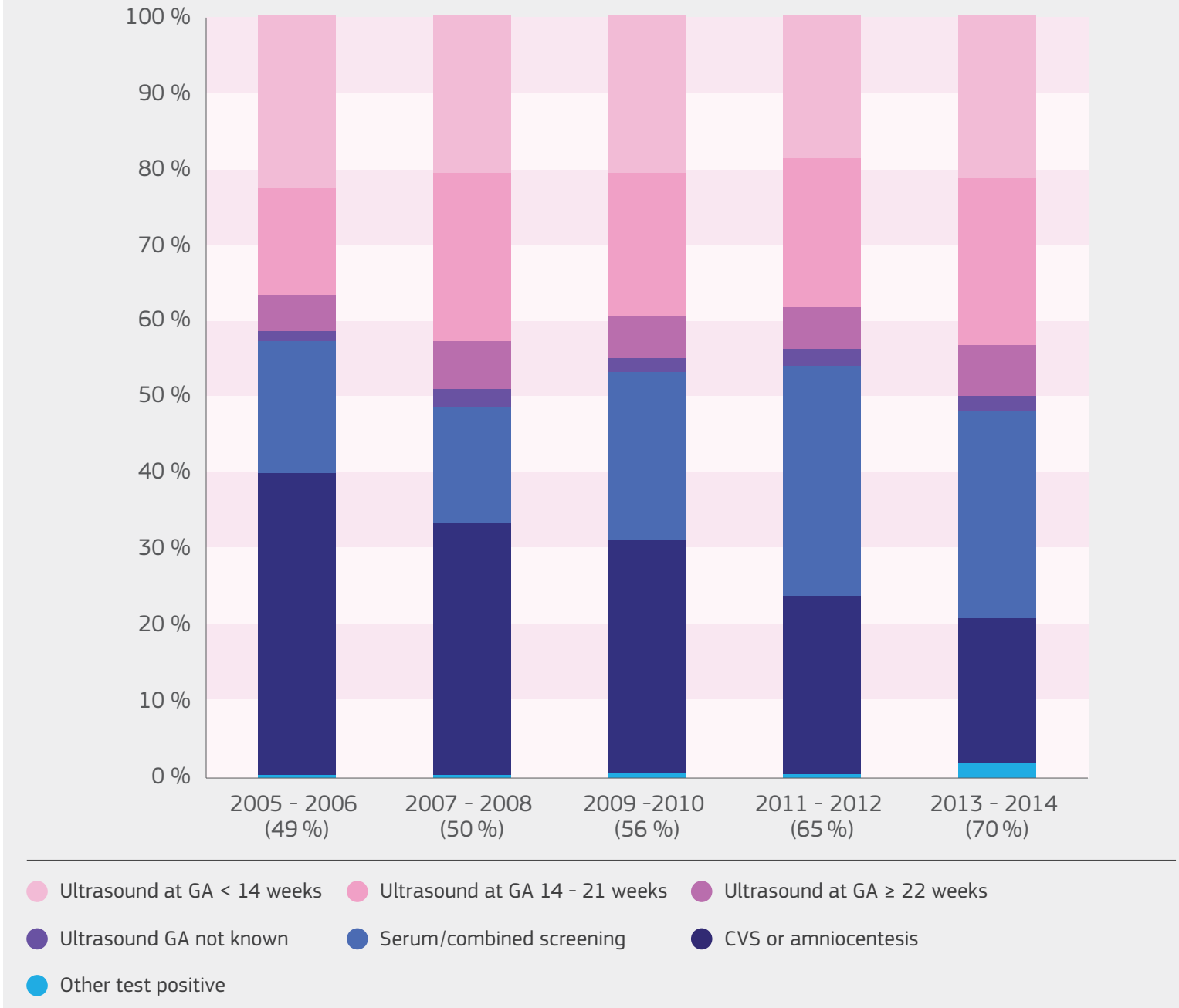


Figure 6. Distribution of the screening techniques reported for prenatal diagnosis of Down syndrome in the EUROCAT registries and % of cases prenatally diagnosed (in brackets). Two-years intervals (2005-2006, 2007-2008, 2009-2010, 2011-2012 and 2013-2014).

Acknowledgements, Acronyms & References

Acknowledgements

We would like to acknowledge all the active and past EUROCAT registries (*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

[1] Morris et al. (2014) Major congenital anomalies in babies born with Down syndrome: a EUROCAT population-based registry study. Am J Med Genet A. 164A:2979-86.

[2] EUROSTAT Fertility statistics:
http://ec.europa.eu/eurostat/statistics-explained/index.php/Fertility_statistics (accessed 31.01.2019)

[3] Khoshnood B (2011) Paper 2: EUROCAT public health indicators for congenital anomalies in Europe. Birth Defects Res A Clin Mol Teratol. 91 Suppl 1:S16-22.

[4] Loane et al. (2013) Twenty-year trends in the prevalence of Down syndrome and other trisomies in Europe: impact of maternal age and prenatal screening. Eur Jo Human Genet, 21:27-33.

Getting in touch with the EU

In person
All over the European Union there are hundreds of Europe Direct information centres. You can find the address of the centre nearest you at: *https://europa.eu/european-union/contact_en*.

On the phone or by email
Europe Direct is a service that answers your questions about the European Union. You can contact this service:
– by freephone: 00 800 6 7 8 9 10 11 (certain operators may charge for these calls),
– at the following standard number: +32 22999696 or
– by email via: *https://europa.eu/european-union/contact_en*.

Finding information about the EU

Online
Information about the European Union in all the official languages of the EU is available on the Europa website at: *https://europa.eu/european-union/index_en*.

EU publications
You can download or order free and priced EU publications at: https://publications.europa.eu/en/publications. Multiple copies of free publications may be obtained by contacting Europe Direct or your local information centre (see *https://europa.eu/european-union/contact_en*).

EU law and related documents
For access to legal information from the EU, including all EU law since 1952 in all the official language versions, go to EUR-Lex at: *http://eur-lex.europa.eu*.

Open data from the EU
The EU Open Data Portal (*http://data.europa.eu/euodp/en*) provides access to datasets from the EU. Data can be downloaded and reused for free, both for commercial and non-commercial purposes.

KJ-BF-19-001-EN-N

The European Commission's science and knowledge service

Joint Research Centre

JRC Mission

As the science and knowledge service of the European Commission, the Joint Research Centre's mission is to support EU policies with independent evidence throughout the whole policy cycle.

EU Science Hub
ec.europa.eu/jrc

@EU_ScienceHub

EU Science Hub - Joint Research Centre

EU Science, Research and Innovation

EU Science Hub

Publications Office

ISBN 978-92-76-00574-2
doi:10.2760/70796