eurocat

european surveillance of congenital anomalies

print **EUROCAT –JOINT ACTION newsletter 2013**

Welcome to the EUROCAT Joint Action Newsletter 2013.

In this last newsletter of the EUROCAT Joint Action we focus on the most important events, documents and publications issued in 2013.

Prevalence Tables

To register for access to EUROCAT's Prevalence and/or Prenatal Detection Rate data tables on the EUROCAT website, click the following link
http://www.eurocat-network.eu/AccessPrevalenceData/PrevalenceTables/Register

The prevalence and risk of Down syndrome in monozygotic and dizygotic multiple pregnancies in Europe: implications for prenatal screening.

Both multiple birth and Down syndrome are associated with older maternal age, but there is very little information available regarding the prevalence of Down syndrome in fetuses and babies from multiple pregnancies compared to singletons. Our objective was to estimate the risk of a fetus from a monozygotic or dizygotic multiple pregnancy being diagnosed with Down syndrome according to the mother's age, and ascertain the likely pregnancy outcome.

Our data came from 10 registries in 8 countries (including specially prepared case data from the NDSCR) and represented almost 15 million births between 1990 and 2009 in Europe; of which 2.88% were multiple births. (full text available at http://onlinelibrary.wiley.com/doi/10.1111/1471-0528.12574/pdf)

Overall we found that individual fetuses from multiple births were at less risk of Down syndrome than singletons with mothers of similar age. In monozygotic pregnancies the risk was approximately one third that of singletons across all maternal ages. In dizygotic pairs, although the risk for each fetus was lower than that of a similar singleton, each fetus has an individual risk and so the risk of at least one fetus from such a pregnancy having Down syndrome was about one third higher than would be expected in a singleton pregnancy.

Fetuses with Down syndrome from multiple pregnancies were less likely to be prenatally diagnosed than singletons at all maternal ages, and those who were prenatally diagnosed were less than half as likely to be induced abortions as singletons with Down syndrome. We found no difference in the rates of still birth between babies from multiple births and singletons with Down syndrome.

Prevalence of selected monogenic syndromes in Europe

Population-based prevalence data on monogenic syndromes are rare because they require coverage of a large population, standardized data collection and genetic expertise. EUROCAT is in a unique position to present data on rare genetic conditions that lack reliable epidemiologic data across Europe. The updated prevalence of selected monogenic syndromes in Europe can be found on EUROCAT website.

Prevalence and clinical manifestations of oculo-auriculo-vertebral spectrum

Oculo-auriculo-vertebral spectrum (OAVS) is a complex developmental disorder characterised by anomalies of the ear, hemifacial microsomia, epibulbar dermoids and vertebral defects. So far epidemiological data were limited and controversial and the causes are believed to be heterogeneous and complex. Using the EUROCAT database we have conducted the largest population-based epidemiological study on OAVS to date. The study included 355 infants with OAVS recorded during the 1990–2009 period in 34 EUROCAT registries active in 16 countries. The prevalence of clinically relevant cases was 3.8 per 100,000 births. Most (95.8%) patients are live born. Of 18.9% diagnosed prenatally, only one-fifth was terminated because of the presence of a severe anomaly/anomalies. Microtia (88.8%), hemifacial microsomia (49.0%) and ear tags (44.4%) were the most frequent anomalies, but a high rate (69.5%) of anomalies of different organ/systems was observed as well. Twinning, assisted reproductive techniques and maternal pre-pregnancy diabetes were confirmed as risk factors.

Barisic I, Odak L, Loane M, Garne E, Wellesley D, Calzolari E, Dolk H, Addor MC, Arriola L, Bergman J, Bianca S, Doray B, Khoshnood B, Klungsoyr K, McDonnell B, Pierini A, Rankin J, Rissmann A, Rounding C, Queisser-Luft A, Scarano G, Tucker D. Prevalence, prenatal diagnosis and clinical features of oculo-auriculo-vertebral spectrum: a registry-based study in Europe. Eur J Hum Genet. 2014 Jan 8. doi: 10.1038/ejhg.2013.287. [Epub ahead of print]

EUROCAT Statistical Monitoring Report 2011

EUROCAT annually performs statistical monitoring for both trends and clusters in time in order to detect signals of new or increasing teratogenic exposures and monitor progress in the prevention of congenital anomalies. The EUROCAT Statistical Monitoring Report 2011 publishes details of the trends and clusters detected in individual registries and in all registries combined (pan-Europe) for the ten year period 2002-2011. Key findings at the European level are:

- Rates of neural tube defects (NTDs) declined on average by 1% per year, with rates for anencephalus declining on average by 2% per year.
- Although there was an overall decreasing trend in congenital heart defects (CHD) over time, increasing trends were detected for some of the more severe types of CHD. Atrioventricular septal defect and tetralogy of Fallot increased on average by 2-3% per year, pulmonary valve atresia increased on average by 4% per year and single ventricle increased on average by 6% per year.
- Prevalence rates for the following digestive anomalies increased on average by 2-4 % per year: ano-rectal atresia and stenosis, duodenal atresia and stenosis, atresia and stenosis of other parts of the small intestine. In contrast atresia of bile ducts decreased by an average of 6% per year.
- The prevalence of craniosynostosis increased on average by 3% per year.
- Increasing trends over time were found for the chromosomal autosomal trisomies (Down syndrome and Edward syndrome), which were explained by the increase in the proportion of older mothers giving birth in Europe. In contrast, prevalence rates of Klinefelter syndrome decreased on average by 4% per year.

Investigation of clusters in the last 2 years (2010-2011) identified no clusters of immediate public health concern. The report also includes the findings of cluster detection conducted at country level i.e. countries with more than one registry are monitored to detect clusters at a national as well as at a regional level. This is the first time this methodology has been applied. The Taskforce for the Evaluation of Clusters (TEC) continues to be available for consultation on clusters identified by statistical monitoring.

Medication use in pregnancy

EUROCAT now has 19 registries providing data on medication use in pregnancy, using the Anatomical Therapeutic Chemical Classification coding system.

During the EUROCAT Joint Action, EUROCAT has conducted some important studies to investigate the teratogenic risk of the use of newer antiepileptic drugs and antidepressants amongst women of childbearing age in order to inform clinical practice and public health interventions.

EUROCAT was able to get important FP7 funding to undertake a project entitled EUROmediCAT – Safety of Medication Use in Pregnancy (www.euromedicat.eu), to further contribute to the development of a European postmarketing surveillance system.

Primary Prevention of Congenital Anomalies

In the framework of the work package 7 of the EUROCAT Joint Action, an online survey was conducted to collect information on the existing health policies and actions related to primary prevention of congenital anomalies (CA) in Europe.

Two questionnaires were developed. The first questionnaire (launched in November 2011) was aimed to provide an update of the current public health measures of primary prevention of neural tube defects (NTD) by raising folic acid (FA) intake and folate status.

The second questionnaire (launched in October 2012) was aimed to collect information on health policies and actions concerning the following CA risk factors: maternal lifestyles; food safety; infectious and chronic diseases; drugs and medications; genetic factors and risk factors related to environment and workplace. **read more**

12th European Symposium on Congenital Anomalies and 28th Annual Registry Leader's Meeting

The **12th international EUROCAT symposium** took place on 14 June 2013 in Zagreb, Croatia and was attended by 186 participants from 26 countries

The main topics of the symposium were marking of the 50th anniversary of the thalidomide tragedy, public health policies in the field of rare diseases, research on genetic and environmental risk factors for congenital anomalies, the use of drugs in pregnancy and evaluation of pre-implantation and prenatal diagnosis. The symposium hosted distinguished keynote speakers from various European countries. There were 19 oral and 57 poster presentations. Abstracts can be found on the EUROCAT website: http://www.eurocat-network.eu/aboutus/publications/eurocatsymposiaandworkshops/zagrebsymposium2013

Participants of the 12th European Symposium on Congenital Anomalies and 28th Registry Leaders Meeting in Zagreb, 14 June 2013

EUROCAT held its **28th Annual Registry Leader's Meeting** from 11-13 June 2013 in Zagreb and it was attended by 85 participants. The representatives from Bulgaria, Lithuania and Saudi Arabia gave presentations on the activities of their registries. We also warmly welcomed our guest, Project and Policy Officer for rare disease, Jaroslaw Waligóra from the European Commission, Directorate General Health and Consumer Protection. Professor Helen Dolk gave a comprehensive overview on the development of EUROCAT activities in the past and future prospects.

EUROCAT Joint Action WP leaders presented on the progress of their work at plenary meetings. Parallel sessions of Folic acid Monitoring Committee, Air pollution Monitoring Committee, Registry Advisory Service, Website Dissemination Committee and EDMP (EUROCAT Data Management Programme) tutorial were held and were also very well attended by many interested participants.

New EUROCAT members

We are very happy to see the EUROCAT membership growing. EUROCAT welcomes new affiliate members: Katya Kovacheva from the Pleven Registry Bulgaria; Ieva Grinfelde and Santa Pildava from Latvia; Flemming Kleist Stenz from the Registry of Congenital Malformations, Greenland and Florence Rouget from the Brittany Registry of Congenital Malformations, France.

We also welcome EUROCAT world affiliates: Dr Rosa Liascovich from the National Registry of Congenital Anomalies of Argentina; Professor Saeed Dastgiri from the Tabriz Registry of Congenital Anomalies; Professor Barry Borman from the Centre for Public Health Research, Wellington, New Zealand and Dr Ahmed M Kurdi from the Prince Sultan Military Medical City, Saudi Arabia.

Other recent publications from EUROCAT

EUROCAT (2014). EUROCAT Special Report: Prevalence of Neural Tube Defects in Younger Mothers in Europe 2000-2008: Analysis of the EUROCAT Database. *EUROCAT Central Registry, University of Ulster*. [Full text]

Barisic I, Odak, L, Loane M, Garne E, Wellesley D, Calzolari E, Dolk H, Addor M-C, Arriola L, Bergman JEH, Bianca S, Boyd P, Draper E, Gatt M, Haeusler M, Khoshnood B, Latos- Bielenska A, McDonnell R, Pierini A, Rankin J, Rissmann A, Queisser-Luft A, Verellen-Dumoulin C, Stone D and Tenconi R (2013). Fraser syndrome: epidemiological study in a European population. *American Journal of Medical Genetics Part A*. 161A: 1012-1018. [Abstract]

CHW Wijers, van Rooij IALM, Bakker M, CLM Marcelis, Addor M-C, Barisic I, Beres J, Bianca S, Bianchi F, Calzolari E, Greenlees R, Lelong N, Latos- Bielenska A, Dias C M, McDonnell R, Mullaney C, Nelen V, O'Mahony M, Queisser-Luft A, Rankin J, Zymak-Zakutnya, N, I de Blaauw, Roeleveld N and de Walle H (2013). Anorectal malformations and pregnancy-related disorders: a registry-based case-control study in 17 European regions. *British Journal of Gynaecology*. [Full text]

EUROCAT and EUROPLAN (2013). Primary Prevention of Congenital Anomalies. *EUROCAT Central Registry, University of Ulster*. [Full text]

Links to other EUROCAT Joint Action news items

For a full list of EUROCAT publications click here

University of ULSTER

Health Programme 2008-2013 Together for Health

Executive Agency for Health and Consumers

Primary Prevention of Congenital Anomalies (continued)

The questionnaires consisted of several items within six main topics:
1) recommendation(s)
2) guidelines
3) training for health professional
4) health educational initiatives, public campaign(s)
5) governmental support, incentives and
6) CA primary prevention plan/strategy laws, regulations, decrees.

22 European countries contributed to the first questionnaire and the second questionnaire was completed by 18 European countries.

Preliminary results show a substantial variation across European countries in developing and implementing recommendations on folic acid and folate intake and on the strategies and activities on primary prevention of CA. Results of the survey will be published in a peer review journal.

The results of this activity have contributed to the development of the EUROCAT (European Surveillance of Congenital Anomalies)/EUROPLAN (European project for Rare Diseases National Plans Developments) recommendations on policies to be considered for the primary prevention of congenital anomalies in National Plans and Strategies on Rare Diseases (http://www.eurocat-network.eu/content/EUROCAT-EUROPLAN-Primary-Preventions-Reccomendations.pdf)

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