

# **EUROCAT Joint Action: Surveillance of Congenital Anomalies**

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*EUROCAT – Odense (Denmark) Registry*

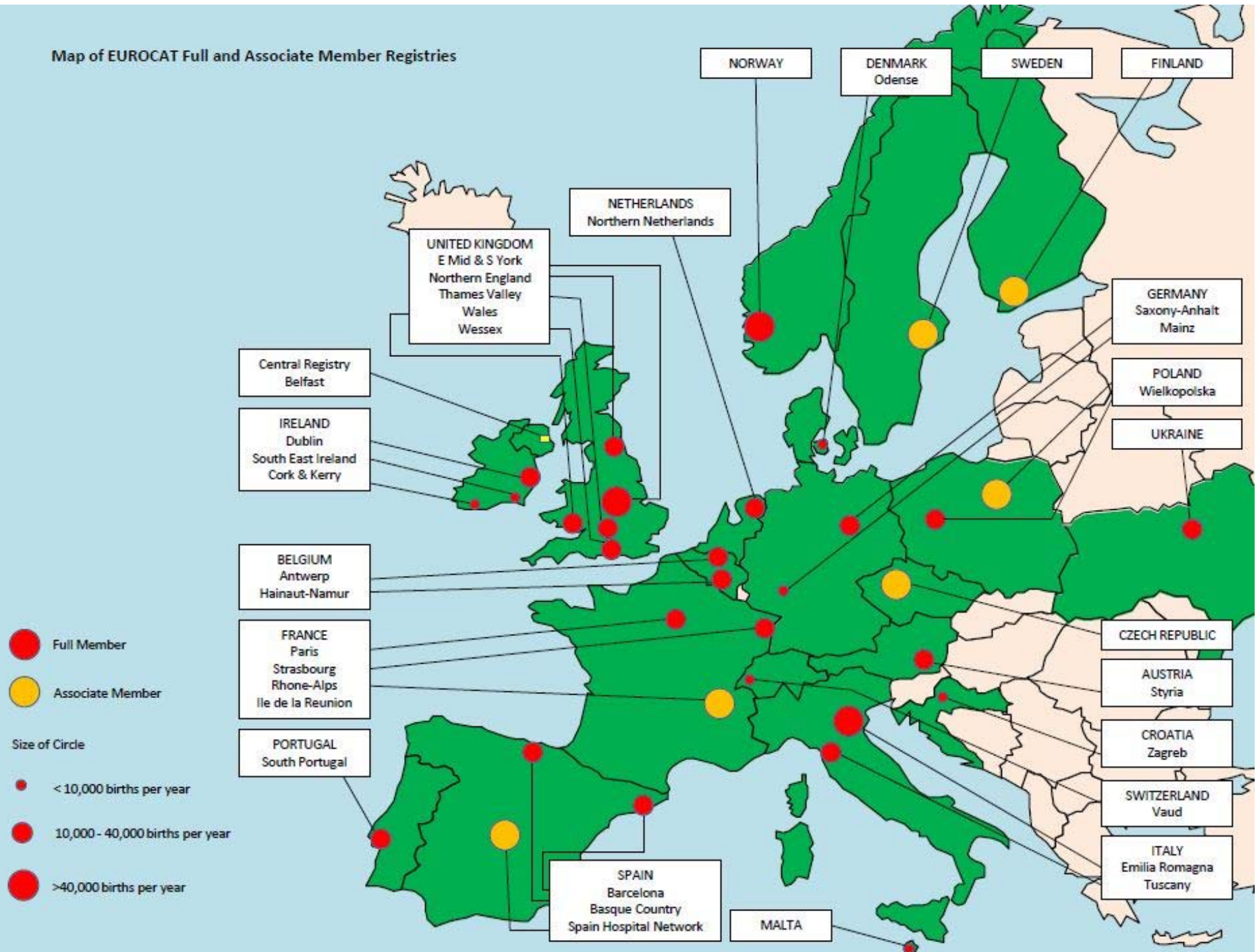
Luxembourg  
25<sup>th</sup>-26<sup>th</sup> October 2011



## About EUROCAT

- **EUROCAT:** The European Surveillance of Congenital Anomalies
- In existence since 1979
- A network comprising almost all of the population-based registries for the epidemiological surveillance of congenital anomalies in Europe
- Currently surveys more than 1.7 million births per year in Europe (31% of birth population covered in the EU via 38 registries in 21 countries)
- Cases of all major structural congenital and chromosomal anomalies among live births, still births and terminations of pregnancy for fetal anomaly following prenatal diagnosis, are registered using multiple sources of information
- Using common software, each member registry transmits either anonymised individual case data (full members) or summary data (associate members) to a central database at EUROCAT Central Registry, where further quality validation is performed

Map of EUROCAT Full and Associate Member Registries



## About EUROCAT

- **General Objective:**
  - Evaluation of the public health impact of congenital anomalies,
  - Report clusters and trends in Europe,
  - Assessment of the teratogenic impact of new or changing environmental exposures
- **EUROCAT Central Registry at University of Ulster - WHO Collaborating Centre for the Surveillance of Congenital Anomalies**
  - To assist the WHO in implementing the resolution WHA63.17 of the 63<sup>rd</sup> World Health Assembly (2010) on birth defects
- **Knowledge**
  - Prevalence of congenital anomalies in Europe with changes over time and differences between countries
  - Coding of congenital anomalies (ICD-11)
- **Care**
  - Primary prevention
- [www.eurocat-network.eu](http://www.eurocat-network.eu)

## ABOUT US

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## Announcements [\[Archive Announcements\]](#)

### EUROCAT as a WHO Collaborating Centre

The Centre for Maternal, Fetal and Infant Research, School of Health Sciences, University of Ulster – home of EUROCAT's Central Registry has been re-designated on 19 May 2011 for a further period of 4 years as a WHO Collaborating Centre for the Surveillance of Congenital Anomalies. In this capacity EUROCAT will assist the WHO in implementing the resolution WHA63.17 of the 63rd World Health Assembly (2010) on birth defects at both a European and global level. EUROCAT will also assist the WHO in implementing its strategy for the prevention and control of non-communicable diseases (NCDs action plan 2008-2013).

## Data from 1980-2009 available on the Website

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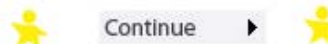
### GALLERY

Where Am I? -> ACCESS PREVALENCE DATA -> [Prevalence Tables](#)

Direct link to this page: <http://www.eurocat-network.eu/ACCESSPREVALENCEDATA/PrevalenceTables>

### Access Prevalence Tables

If you would like to interrogate the database further  
to your own specifications, please



Some example tables:

PDF Version [Table "Prevalence 96 Subgroups Last 5 years"](#)

PDF Version [Table "Down Syndrome Last 5 Years"](#)

The results generated here are in PDF format and you will need an appropriate application installed to view them.  
You can click on the icon below to install the Adobe Reader.



## EUROCAT Prevalence Data Tables

A1 - cases and prevalence (per 10,000 births) for the following registries: All Full Member Registries, from 2005 - 2009

Anomaly	LB N	FD N	TOPFA N	LB+FD+TOPFA N	LB+FD+TOPFA Rate	Excluding Chromosomal	
						LB+FD+TOPFA N	LB+FD+TOPFA Rate
<b>All Anomalies</b>	60621	1644	13863	76128	232.21	64071	195.43
<b>Nervous system</b>	3707	359	4023	8089	24.67	7326	22.35
Neural Tube Defects	784	152	2355	3291	10.04	3148	9.60
Anencephalus and similar	90	87	1080	1257	3.83	1228	3.75
Encephalocele	106	20	267	393	1.20	372	1.13
Spina Bifida	588	45	1008	1641	5.01	1548	4.72
Hydrocephaly	941	98	862	1901	5.80	1720	5.25
Microcephaly	716	43	68	827	2.53	755	2.31
Arhinencephaly/holoprosencephaly	92	28	329	449	1.37	302	0.92
<b>Eye</b>	1096	11	86	1193	3.64	1081	3.30
Anophthalmos/microphthalmos	258	5	54	317	0.97	260	0.79
Anophthalmos	48	2	19	69	0.21	64	0.20
Congenital cataract	334	0	4	338	1.03	326	0.99
Congenital glaucoma	93	0	0	93	0.28	91	0.28

## Most types of congenital anomalies are rare diseases

### 2005-2009 data from 31 full member EUROCAT registries:

67 of EUROCAT's 95 standard congenital anomaly subgroups have a live birth prevalence of <5 per 10,000 births

Examples (live birth rate per 10,000 births):

Spina Bifida:	1.89
Hydrocephaly:	2.99
Hypoplastic left heart:	1.48
Atresia of bile ducts:	0.25
Achondroplasia:	0.28
Conjoined twins:	0.03
Trisomy 13:	0.38

## How many cases in Europe?

- Combined livebirth prevalence of the 67 subgroups is 61.39 per 10,000 births which equates to
  - 30,695 affected live births across Europe per year
- Live birth prevalence of chromosomal anomalies is 15.01 per 10,000 births:
  - 7505 affected live births across Europe per year
- Live birth prevalence of single gene syndromes diagnosed in infancy is 4.79 per 10,000 births
  - 2395 affected live births across Europe per year  
(minimum number due to coding problems of very rare syndromes)

## Levels of Prevention of Congenital Anomalies

### 1. Primary Prevention:

- pre disease (attacking basic cause(s) of disease, altering environment (examples later)
- organogenesis is early in pregnancy often before pregnancy is confirmed
- Primary prevention needs to start preconceptionally

### 2. Secondary Prevention:

- disease has started but symptoms have not appeared (detecting and treating early to prevent disease development) e.g. better outcomes after surgery for severe congenital heart defects if diagnosed prenatally

### 3. Tertiary Prevention:

- Disease has become symptomatic (curing, controlling or preventing complications) e.g. paediatric surgery

## Methods of Primary Prevention of Congenital anomalies

Avoidance of exposure to known teratogenic agents

Cessation of maternal smoking, alcohol consumption and recreational drugs

Maternal infection control – e.g. continuation of the rubella vaccination programme

Maternal nutritional considerations (general healthy diet, periconceptual use of folic acid, vitamin B12 status, avoid excess of vitamin A, reduce deprivation)

Appropriate clinical care (particularly in the periconceptual period) of high risk women with chronic diseases (diabetes, epilepsy, depression) who needs to continue drug treatment

Pre-marketing drug testing, post-marketing pharmacovigilance of drugs taken during pregnancy (of unknown teratogenicity), health technology surveillance

Reduction of exposure to environmental pollutants (envirovigilance)

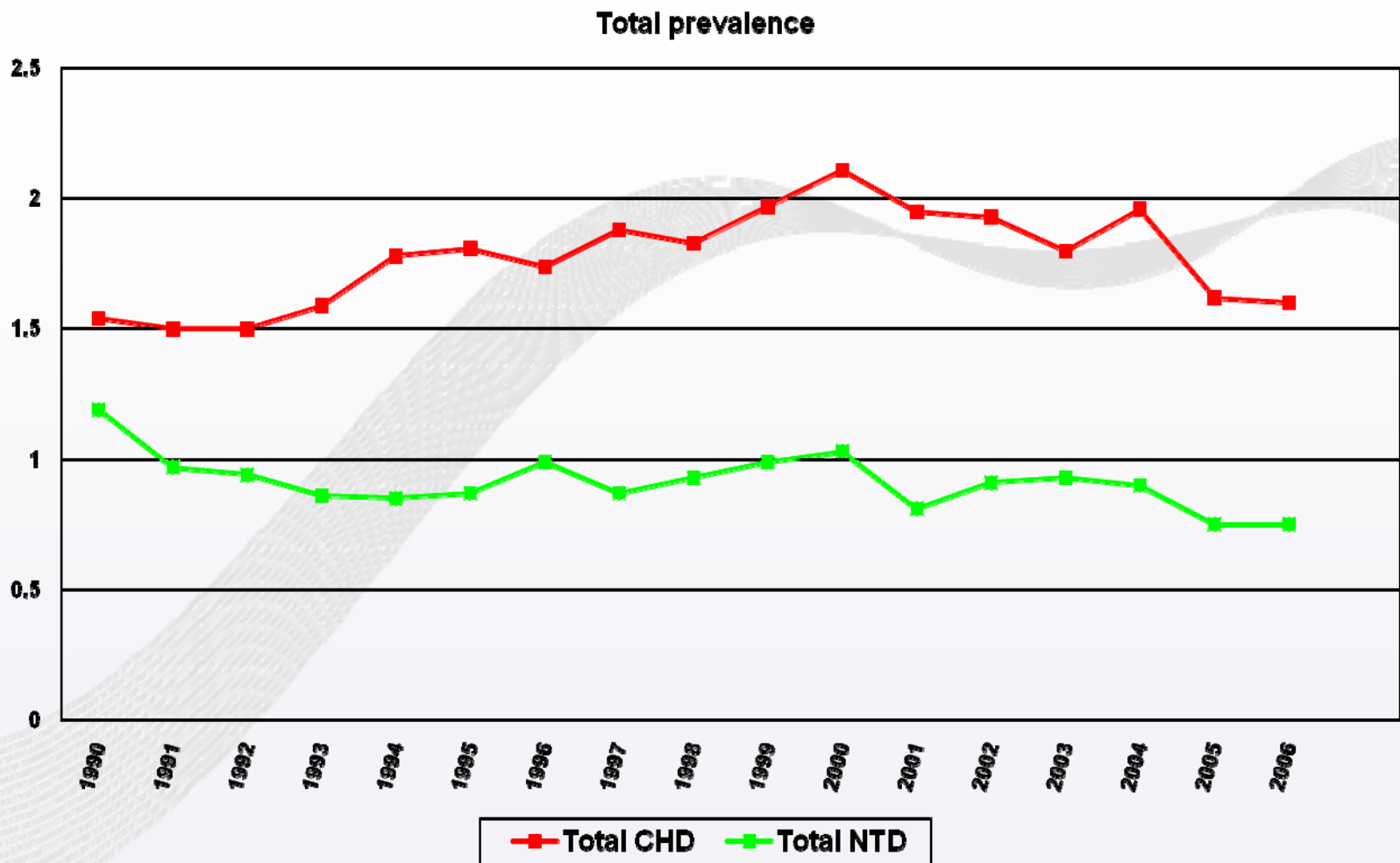
Genetic counselling for high risk families

**TO BE INTEGRATED INTO NATIONAL RARE DISEASE PLANS (LINKS WITH EUROPLAN)**

## Pan-European trends in total prevalence – 10 years

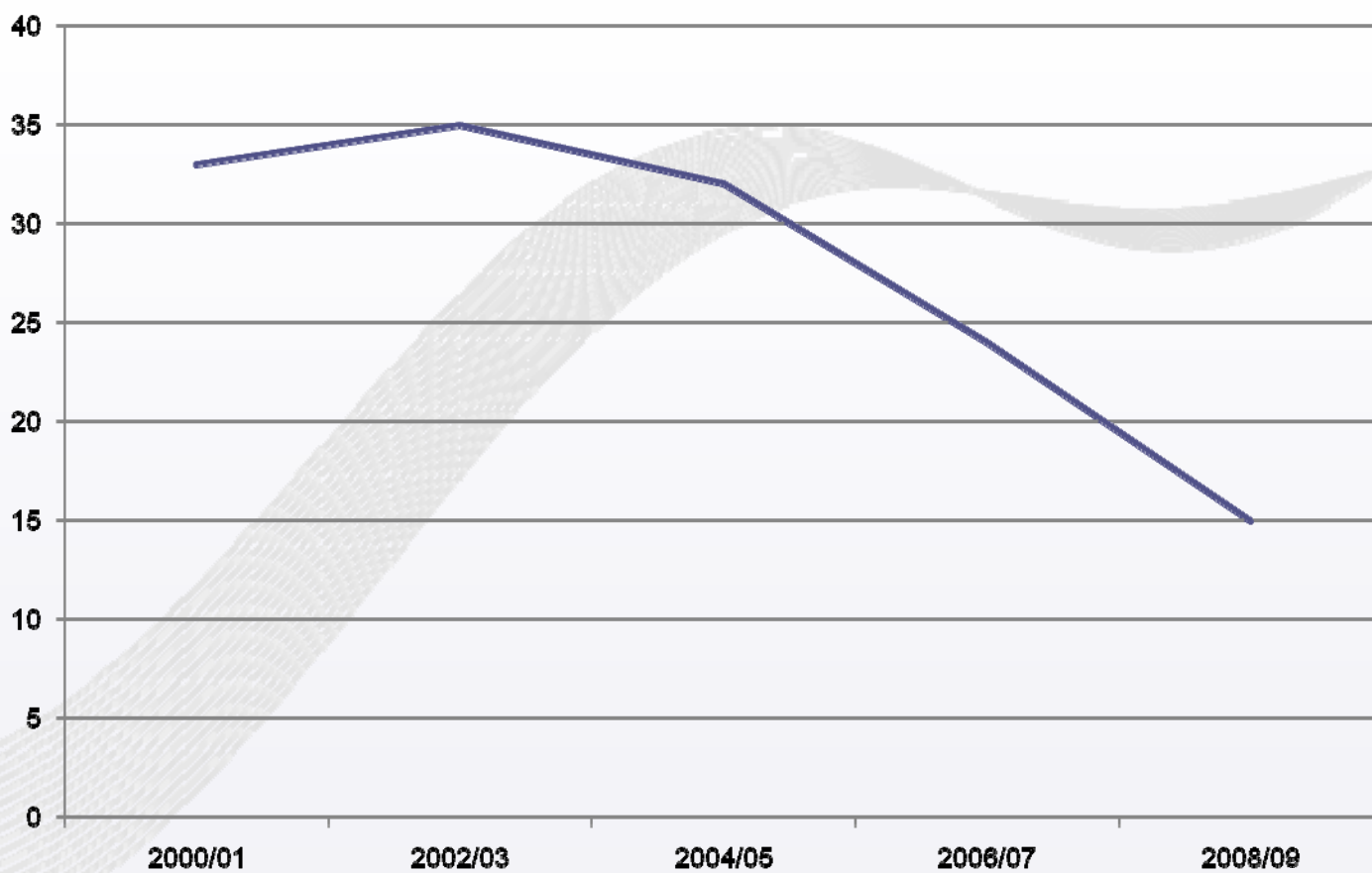
- **Increasing trends**
  - Gastroschisis
  - Hypospadias
  - Trisomy 13, 18
  - Trisomy 21
- **Decreasing trends**
  - Severe congenital heart defects
  - Neural tube defects
  - Atresia of bile duct
  - Bilateral renal agenesis

## Decreasing trends of severe cardiac defects (13 subgroups) and neural tube defects 1990-2006



## Biliary atresia EUROCAT 2000-2009

Number of cases



$P < 0.001$

## Structure of EUROCAT 2011-2013

Central EUROCAT registry at University of Ulster, Belfast

### 9 Workpages

WP1 - Coordination (Helen Dolk, UK)

WP2 - Dissemination (Ingeborg Barisic, Croatia)

WP3 - Evaluation (Helen Dolk, UK)

WP4 - Registration, central database and surveillance (Maria Loane, UK)

WP5 - Coding and classification, and data quality (Ester Garne, Denmark)

WP6 - Investigation of trends, clusters and new exposures (Martine Vrijheid, Spain)

WP7 - Primary prevention (Domenica Taruscio, Italy)

WP8 - Prenatal screening, Down Syndrome, and genetic syndromes (Joan Morris, UK)

WP9 - Medication during pregnancy (Marian Bakker, The Netherlands)

## For Further Information

**Prof. Helen Dolk (Project Leader)**  
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## EUROCAT Joint Action 2011-2013

- EUROCAT is currently funded as a Joint Action of the EU and Member States through the DG Sanco Public Health Programme
- **Strategic Relevance:** Congenital anomalies are a major group of mainly rare diseases where concerted action across Europe has been identified as a priority in the Council Recommendation of 8th June 2009 on an action in the field of rare diseases, and in the Communication from the Commission on Rare Diseases: Europe's challenges of November 2008
- These recognise the need for registries and databases co-ordinated at European level, for pooling of expertise, improving the coding and classification of rare diseases, for comparable epidemiological data at EU level, and for identifying the possibilities for primary preventive measures
- The Joint Action combines funding of the EU and Member States in order to secure a sustainable, high quality and easily accessible information system on congenital anomalies for almost one third of the European birth population
- Through the Joint Action EUROCAT expects to have an important impact on future Member States policy on rare diseases