Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

Congenital Anomalies Cork & Kerry

Volume 3, Issue 1, 2010

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EUROCAT Special Report: The Status of Health in the European Union: Congenital Malformations (June 2009)

http://www.eurocat.ulster.ac.uk/pdf/Special-Report-Con-Malfs.pdf

Trends in congenital anomalies are reviewed in a recent EUROCAT Special Report. There has been a slight increase in the prevalence of all congenital anomalies since the mid-1990s. This increase has been due in part to an increase in congenital heart disease (CHD). Improvements in data quality and increases in risk factors contribute. Prevalence of Down syndrome has increased due to the increase in the average maternal age in Europe. A strong increase in gastroschisis prevalence has occurred in Europe and elsewhere; the reasons for this are not fully understood.

Prenatal screening, and the proportion of cases diagnosed prenatally, varies in Europe. Prenatal diagnosis can improve survival in some cases.

The cause of the majority of cases of congenital anomaly is unknown but is suspected to result from a combination of environmental and genetic factors. A chromosomal abnormality is found in 15% of cases, in addition <5% can be attributed to a known single gene mutation and another <5% to exposure to a single environmental teratogen (such as drug taken during early pregnancy). Studies looking at socioeconomic differences have shown the importance of environmental factors. Genetic susceptibility to these factors is likely to vary in the population.

- Low **folic acid** status in peri-conceptual period is an established risk factor for neural tube defects (NTD). The fortification of a staple food with folic acid raises the preconception folate status of women and is a proven successful strategy to prevent NTD. The current approach, to recommend peri-conceptual folic acid supplementation is not effective.
- **Epilepsy** and **diabetes** are associated with an increased risk of congenital anomalies. Appropriate treatment can reduce this risk.
- **Rubella** vaccination prevents congenital rubella syndrome
- **Smoking**, **alcohol**, and **recreational drug** (cocaine/solvent) use are harmful to the foetus, particularly in early pregnancy.
- Poor **nutritional status** may coexist and add to the risk from other risk factors
- **Assisted reproductive technology** (ART) has increased. There are concerns about congenital anomaly risk with the use of ART. Strict confidentiality in relation to ART has made it a difficult area to research.
- The evidence regarding risk associated with **other chemicals** is poor. A precautionary approach is recommended.

The total prevalence of major congenital anomalies recorded by EUROCAT between 2000 and 2004 was 23.8 per 1,000 births (19.9 per 1,000 live births). The most commonly recorded anomalies were: congenital heart disease (6.4/1,000 births); chromosomal abnormalities (3.36/1,000 births); and limb defects (3.6/1,000 births). The most common chromosomal abnormality was Down syndrome (1.92/1,000 births).

Cork and Kerry Congenital Anomaly Registry: 2004 and 2005 data

2004: There were 8555 live births and 63 stillbirths born in Cork & Kerry in 2004. The sex ratio male : female at birth was 1.07:1. Almost three percent (2.9%) of births, 251 babies with a birth defect, were registered in 2004 on the Cork and Kerry Congenital Anomaly Registry (243 singleton and 8 twin deliveries). These comprised 236 livebirths and 15 stillbirths. 29 cases (11.5%) did not survive beyond one week of age. There were 132 male and 118 female infants born with a birth defect with 1 infant of indeterminate sex. The age range of mothers who gave birth to a child with a congenital anomaly in 2004 was 17 to 48 years. Three quarters (76%) of congenital anomalies were diagnosed within the first week of life.

2005: There were 8621 live births and 42 stillbirths in Cork & Kerry in 2005. The sex ratio of births was 1 in 2005. 225 babies with a birth defect, 2.6% of births, were registered in the Cork and Kerry Congenital Anomaly Registry in 2005 - 217 singleton and 8 twin deliveries. 218 were live births and 7 stillbirths. 17 cases (7.6%) did not survive beyond one week of age. There were 112 males and 113 female infants born with a birth defect. The age range of mothers who gave birth to a child with a congenital anomaly in 2005 was 17 to 43 years. 81% were diagnosed within the first week of life. Babies born with a congenital anomaly are significantly more likely to be born prematurely and to be of low birth weight. In 2005, 20% were under 37 weeks gestation and 18% were under 2500g weight compared with 5% respectively of all births nationally (χ^2 p <0.0001).

Risk factors Information was available on maternal smoking and alcohol consumption during pregnancy for 82% of mothers. 69% did not drink or smoke during pregnancy, 20% smoked, 8% drank and 3% drank and smoked. The use of folic acid was added to the database as a separate variable in 2005 and was recorded in 63% of cases in 2005. Where known 15% did not take folic acid, 50% took pre-conceptual folic acid and a further 35% took post-conceptual folic acid. Table 1 shows the breakdown by anomaly by year for babies born in Cork and Kerry in 2004 and 2005.
The prevalence of congenital anomaly increases with maternal age with the rate /1000 increased in the 40-44 age group. Table 2).

Table 1. Cases of congenital anomaly and prevalence per 1,000 births from Cork and Kerry Registry data , 2004 and 2005 (includes Live Births, Foetal Deaths and Terminations of Pregnancy where data available)

	2004		2005	
Congenital Anomaly	Cork & Kerry cases	Cork and Kerry prevalence	Cork and Kerry cases	Cork and Kerry prevalence
All cases	251	29.13	225	25.973
Anomaly				
Nervous System	25	2.90	22	2.54
Eye	1	0.12	4	0.462
Ear, Face & Neck	1	0.12	2	0.231
Congenital Heart Disease	64	7.43	63	7.272
Respiratory	4	0.46	3	0.346
Oro-facial clefts	11	1.28	21	2.424
Digestive system	14	1.63	17	1.962
Abdominal wall defects	6	0.70	4	0.462
Urinary	18	2.09	12	1.385
Genital	21	2.44	29	3.348
Limb	61	7.08	66	7.619
Musculo-skeletal	9	1.04	4	0.462
Other malformations	4	0.46	6	0.693
Teratogenic syndromes with malformation	0	0	1	0.115
Genetic syndromes & microdeletions	6	0.70	12	1.385
Chromosomal	25	2.90	36	4.156

Table 2. Total number of live-births, live-and still-births with congenital anomalies and rate of live- and still-births per 1000 live-births, Cork and Kerry 2004-2005

Age Group of Mother	*Live Births (Cork & Kerry)	Births with congenital anomalies (Cork & Kerry)	Rate Livebirths & Still-births/1000 Livebirths
<20	577	15	26
20-24	2028	55	27
25-29	3788	87	23
30-34	6244	181	29
35-39	3815	107	28
40-44	681	28	41
>=45	22	2	90
Not known	21	1	NA
Total	**17176**	**476**	**28**

The Eurocat prevalence rate based on Eurocat Full Registry Data was 24.74 cases of congenital anomaly per 1000 births (includes live births, foetal deaths and terminations of pregnancy where data available) in 2004 and the rate for 2005 was 24.79. There is more variability in prevalence rate from year to year where small numbers are involved as in the individual Cork and Kerry Register. In addition completeness of data collection varies between registries. EUROCAT-Central Registry reviews Data Quality Indicators by register.

EUROCAT Special Report: Congenital Heart Defects in Europe, 2000-2005 (March 2009)

It is estimated that about 400 babies with congenital heart disease are born each year in Ireland. EUROCAT recently published a report on congenital heart defects in Europe 2000-2005. Congenital heart disease (CHD) accounts for nearly one third of major congenital anomalies diagnosed prenatally or in infancy in babies in Europe. CHD is a diverse group of conditions in terms of severity and associated morbidity and mortality. Improved prenatal diagnosis may have the potential to improve early treatment success further in Europe.

Prevalence

- The average total prevalence rate for CHD in Europe was 8 per 1,000 births, including live births, stillbirths and terminations of pregnancy for foetal anomaly (TOPFA).
- Most cases of congenital heart disease (88%) are non-chromosomal.
- Of these, 75% involve ventricular septal defect (VSD), atrial septal defect (ASD) or pulmonary value stenosis (PVS). The vast majority are live born and survive the first week of life. Less than 10% require surgery. Most survive to adulthood.
- The remaining 25% mainly include conditions with higher perinatal mortality and high (~ 80%) surgery rates. Seven percent are too severe for surgery. Termination of pregnancy is the outcome for a significant proportion of severe cases in some countries.
- 18% of CHD cases had multiple malformations of different organ systems, a similar proportion regardless of CHD severity category.
- 12% of congenital heart disease is associated with chromosomal anomalies (7% Downs's syndrome, 1% Trisomy 13 and 2% Trisomy 18). Chromosomal anomalies are particularly common among atrioventricular septal defects (57% chromosomal).
- Children with CHD are at higher risk of intellectual, sensory and motor impairments.

Diagnosis

- The majority of cases of CHD are diagnosed before the child is one month. Some cases of VSD/ASD/PVS, and aortic valve atresia/stenosis are diagnosed later.

Mortality

- The rate of first week death is 2.7% of all live births with CHD. Another 6% will die within the first year of life.
- Perinatal mortality is high for single ventricle (26.7%), Ebsteins anomaly (19.8%) and Hypoplastic Left Heart (17.2%).

Trends

- The prevalence of CHD has not declined over time. Improved diagnosis of small muscular defects with early spontaneous closure, diagnosed neonatally, has led to an increase in the reported prevalence of CHD

Ireland 2000-2005

- In Ireland the prevalence of CHD between 2000-2005 was 6.62 per 1,000 births (Irish EUROCAT Registry data).

Cork and Kerry in 2004 and 2005

- 128 cases of CHD in total in 2004 and 2005 (see table 1)
  - 101 cases had non-chromosomal CHD. Of these 82 were isolated CHD, 41 being single isolated cardiac cases
  - 18 infant deaths were recorded, 11occurred in the perinatal period.
  - Information on surgery was gathered in 2005 only. Of the 63 cases in 2005, 24 (38%) did not require surgery. One third were scheduled for surgery in infancy. Four cases (6%) were deemed too severe for surgery. Information on surgery was not known for 21% of cases.

Table 1. Number and prevalence per 1,000 live-births of all CHD and non-chromosomal CHD in Cork and Kerry, 2004 and 2005

	2004				2005			
	All CHD	Prevalence per1,000 births	Non-chromosomal CHD	Prevalence per1,000 births	All CHD	Prevalence per 1,000 births	Non-chromosomal CHD	Prevalence per1,000 births
VSD/ASD/PVS[1]	50	5.80	45	5.22	55	6.35	39	4.50
Severe CHD[2]	18	2.09	13	1.51	21	2.42	15	1.73
Very Severe CHD[3]	2	0.23	2	0.23	3	0.35	3	0.35
Total cases	**65**	**7.54**	**56**	**6.50**	**63**	**7.27**	**45**	**5.20**

1. Ventricular septal defect, atrial septal defect, pulmonary valve stenosis
2. Pulmonary valve atresia, Common arterial truncus, Atrioventricular septal defect, Aortic valve atresia/stenosis, Transposition of great vessels, Tetralogy of Fallot, Total anomalous pulmonary venous, Coarctation of aorta
3. Single ventricle, Hypoplastic left heart, Hypoplastic right heart, Ebstein's anomaly, Tricuspid atresia

References

EUROCAT Special Report CDH http://www.eurocat.ulster.ac.uk/pdf/ML/Reports/EUROCAT-Special-Report-CHD.pdf

Risks for a variety of different birth defects are increased after early pregnancy use of the antiepileptic drug valproic acid.

A study by the EUROCAT-Antiepileptic-Study Working Group published in the New England Journal of Medicine June 2010 reports the risks of specific birth defects after first trimester valproic acid use. Exposure in the first trimester has the potential to cause birth defects because this is the period when the major organs are developing. The study used a dataset including 98,075 live births, stillbirths, or terminations with malformations from a population of 3.8 million births in 14 European countries 1995-2005. A significantly increased risk was reported for a variety of specific birth defects with first trimester exposure to valproic acid compared with no antiepileptic drug use. The risk of having a baby with one of the following birth defects - spina bifida, atrial septal defect, cleft palate, hypospadias, polydactyly, craniosynostosis and limb reduction was 2-12 times greater for mothers exposed to valproic acid during pregnancy compared to mothers with no antiepileptic drug use during pregnancy.

The study also compared the use of valproic acid with the use of other antiepileptic drugs and found similar results, suggesting that valproic acid has higher risks associated with birth defects compared to other antiepileptic drugs. Although the relative risks of several birth defects were increased after 1st trimester exposure of valproic acid, it should be recognised that the risks of these specific birth defects are still low, ranging from 1 to 7 per 1,000 exposed pregnancies.

The Cork and Kerry Congenital Anomaly Register collects and registers information on all babies with a birth defect born to mothers resident in Cork and Kerry whether stillborn or live born. The registry is a member of the EUROCAT registries, an established European wide network of congenital anomalies registries. http://www.eurocat-network.eu/ Breaks in staffing have delayed data collection.