

Lenus: Research Repository



Congenital Anomalies Cork & Kerry Newsletter; Vol 11 (1), 2022

Item Type	Other
Authors	Health Service Executive (HSE) South (Cork & Kerry) Department of Public Health;Public Health Area D (Cork & Kerry)
Publisher	Health Service Executive (HSE) South (Cork & Kerry) Public Health Area D Department of Public Health
Rights	Attribution-NonCommercial-NoDerivatives 4.0 International
Download date	2026-04-19 05:43:15
Item License	https://creativecommons.org/licenses/by/4.0/
Link to Item	https://hdl.handle.net/10147/635465



Congenital Anomalies Cork & Kerry

Volume 11 Issue 1, 2022



In this Issue:

- Recent Literature Recommendations
- Birth Defect Surveillance and Clubfoot Online Courses for Healthcare Workers
- Scottish experience of establishing a National Congenital Anomaly Registry
- Some Useful links for further information on specific anomalies detected during pregnancy
- Epidemiology of Cork & Kerry Congenital Anomaly Register Data 2020

References available on request

Cork & Kerry Congenital Anomaly Registry

Public Health Area D
(Cork & Kerry),
Health Service Executive,
Floor 2, Block 8,
St. Finbarr's Hospital,
Douglas Road, Cork.
T12 XH60
Tel: 021 4927601
Fax: 021 4923257
Email: dph@hse.ie

Website:

<http://www.hse.ie/congenitalanomalyregistersireland>

<http://www.eurocat-network.eu/>

Editorial Staff:

Dr Mary O'Mahony,
Consultant in Public Health Medicine
Prof Patricia Kearney
Specialist Registrar Public Health
Medicine
Dr Catherine Crowe
Specialist Registrar Public Health
Medicine
Maria Ryan
Registry Research Nurse
Judy Cronin
Health Informatics Manager

Recent Literature Recommendations

A global update on the status of prevention of folic acid-preventable spina bifida and anencephaly in year 2020: 30-Year anniversary of gaining knowledge about folic acid's prevention potential for neural tube defects

Kancherla V, Wagh K, Priyadarshini P, Pachón H, Oakley GP Jr.
Birth Defects Res. 2022 Nov 7. doi: 10.1002/bdr2.2115. Epub ahead of print. PMID: 36345648.
<https://onlinelibrary.wiley.com/doi/10.1002/bdr2.2115>

The “analysis showed that 61,680 Folic Acid Preventable Spina Bifida and Anencephaly (FAB SBA) cases were prevented in the year 2020 through mandatory folic acid fortification of cereal grains in 58 countries, translating to 22% prevention of total possible FAP SBA prevention globally. Many countries in Africa, Asia, and Europe are yet to implement fortification. In 2020, 30 years after the MRC study was published, 218,270 preventable FAP SBA cases still occurred globally.”

The DoH Folic Acid WG –(2019) <https://assets.gov.ie/228532/293d8869-a5ed-4a2a-9a59-3417f18958a9.pdf> The Committee considers that there is considerable evidence in support of the introduction of mandatory fortification of foodstuffs with folic acid (FA). However, it also recognises that this would be a significant undertaking in terms of resources and timing.” “Committee conclusions on neural tube defects in Ireland: No significant progress has been made in reducing the incidence of NTDs in Ireland in the last decade. NTDs remain highly significant in Ireland in terms of pregnancy loss and infant death, lifelong disability and the wellbeing of parents and families. NTDs are consistently most likely to affect unplanned pregnancies, lower socio-economic families and women who have already had a baby previously with a NTD. Accurate and timely information on incidence of NTDs is critical to inform future measures.”

Information needs of parents of children with congenital anomalies across Europe: a EUROLINKCAT survey

Marcus E, Latos-Bielenska A, Jamry-Dziurla A, Barišić I, Cavero-Carbonell C, Den Hond E, Garne E, Genard L, Santos AJ, Lutke L, Matias Dias C, Neergaard Pedersen C, Neville AJ, Niemann A, Odak L, Pierini A, Rico J, Rissmann A, Rankin J, Morris JK. BMC Pediatr. 2022 Nov 12;22(1):657. doi: 10.1186/s12887-022-03734-z. PMID: 36368959; PMCID: PMC9652126.
<https://bmcpediatr.biomedcentral.com/articles/10.1186/s12887-022-03734-z>

Birth Defect Surveillance and Clubfoot Online courses for Healthcare Workers

Online Self-Paced Course on Birth Defect Surveillance and Prevention

The online course remains available for interested professionals. The course has been developed by the International Centre on Birth Defects (ICBD Centre) and supported in part by funding from the National Center on Birth Defects and Developmental Disabilities, US Centers for Disease Control and Prevention, through Agreement with the Task Force for Global Health. The course is designed for clinicians, epidemiologists, public health professionals, and anyone interested in understanding birth defects and improving their prevention and care. The course includes videos, quizzes, a discussion forum, and publications/resources. It is available in English and Spanish. Additional information is available at <http://www.icbdsr.org/online-self-paced-course-on-birth-defect-surveillance-and-prevention/>.

Please, register through the online [registration form](#).



Clubfoot can be treated: A Course for Healthcare Workers

This course is targeted at healthcare workers and other professionals who care for babies and young children in their work. Its purpose is to raise awareness of the prevalence and impact of birth impairments, and the need to start the appropriate treatment as early as possible and support families who are affected. It takes a close look at clubfoot as one of several birth impairments which require early detection and treatment. It presents essential information on clubfoot and the non-surgical

Ponseti method available to eliminate this lifetime disability.

Information in English: <https://bit.ly/PAHOVCClubfoot>,

Organized by PAHO-WHO, Virtual Campus for Public Health, and MiracleFeet

Scottish experience of establishing a National Congenital Anomaly Registry



Population of Scotland – approx. 5 million with 50,000 births annually.

~1,300 affected babies born with a congenital anomaly annually

<https://publichealthscotland.scot/media/9487/2021-10-05-congenitalanomaliesinscotland2019-table-1-prevalence.xlsx>

The national surveillance of congenital anomalies has long been a goal that is yet to be realised for Ireland. Scotland has recently commenced national surveillance of congenital anomalies, for a population of similar size to Ireland and with modest additional resource. This is made possible in Scotland by enabling legislation, coupled with routine use of a unique health identifier for all patient healthcare contacts. We outline below what we have learned from Scotland on how they achieved national surveillance and in record time.

CARDRISS

CARDRISS is the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland managed by Public Health Scotland (PHS). It is managed by a Consultant in Public Health Medicine, Public Health Scotland- Dr Rachel Woods and supported by a small team of 3 WTE staff. Working in collaboration with the Scottish National Cancer Register, 3 additional Cancer Registration personnel form a cancer and congenital anomaly registration team with personnel accessing the full medical records, ensuring the complete validation and surveillance of babies born with a congenital anomaly are registered on the system. **CARDRISS is a national register for congenital anomalies in Scotland (established in 2021).** It collects and holds information about babies <12mths in Scotland with a major structural or chromosomal condition or recognised syndrome, babies with a condition that ends in a stillbirth or miscarriage (*from 20 weeks*) or terminations of pregnancy (*at any stage*) are also included. Existing Scottish national medical record databases facilitated case ascertainment to meet JRC-EUROCAT associate registry criteria. Consent is not required from families for entry onto the registry by clinicians.

CARDRISS is the Congenital Conditions and Rare Diseases Registration and Information Service for Scotland.

CARDRISS collects and holds information about babies in Scotland with a congenital condition, such as cleft lip and palate, congenital heart disease, or Down syndrome (trisomy 21).

CARDRISS uses this information to help the NHS to:

- monitor trends in specific conditions
- plan and improve services for affected families
- monitor pregnancy and newborn screening programmes
- support research

For further information about CARDRISS, including more on how we use personal information, visit www.publichealthscotland.scot/cardriss

NHS SCOTLAND

Public Health Scotland

CARDRISS is the Congenital Conditions and Rare Diseases Registration & Information Service for Scotland managed by Public Health Scotland (PHS). It is managed by a Consultant in Public Health Medicine, Public Health Scotland- Dr Rachel Woods and supported by a small team of 3 WTE staff. Working in collaboration with the Scottish National Cancer Register, 3 additional Cancer Registration personnel form a cancer and congenital anomaly registration team with personnel accessing the full medical records, ensuring the complete validation and surveillance of babies born with a congenital anomaly are registered on the system. **CARDRISS is a national register for congenital anomalies in Scotland (established in 2021).** It collects and holds information about babies <12mths in Scotland with a major structural or chromosomal condition or recognised syndrome, babies with a condition that ends in a stillbirth or miscarriage (*from 20 weeks*) or terminations of pregnancy (*at any stage*) are also included. Existing Scottish national medical record databases facilitated case ascertainment to meet JRC-EUROCAT associate registry criteria. Consent is not required from families for entry onto the registry by clinicians.

See link to the [CARDRISS patient Information leaflet](#).

Public Health Scotland has a statutory obligation to collect national health data to inform health policy. Scottish legislation recognises Public Health Scotland in law as a producer of official statistics and therefore may, where appropriate, apply the exemptions that exist in the Data Protection Act 2018 that enable it to achieve its purposes in relation to this legal basis for processing. Statistics inform a vast range of decisions across society.

Public Health Scotland holds NHS health and health related data for over 5 million people in Scotland, which in some cases span an individual's whole life from before birth, with the mother's antenatal records, through to that individual's death registration record. This is a wealth of information which can be linked, summarised and analysed to support research studies. See the National Data Catalogue details database held by Public Health Scotland.

<https://www.ndc.scot.nhs.uk/National-Datasets/Full-A-Z/index.asp>

CARDRISS helps Public Health Scotland to fulfil its purpose by providing information on and promoting the health of babies with congenital conditions.

Access to these databases facilitated the publication of annual national statistics on congenital anomalies for the region. See Congenital Anomalies in Scotland (2000-2019), published Oct 2021

<file:///Z:/My%20Documents/DRMOMAHO/Congenital%20Anomaly%20Register/Scotland%20Registry/2021-10-05-congenitalanomaliesinscotland2019-main-report.pdf>

and the Summary report

<https://publichealthscotland.scot/media/9502/2021-10-05-congenitalanomaliesinscotland2019-summary.pdf>

Some Useful links for further information on specific anomalies detected during pregnancy– see also

MyChild.ie

NORD: <https://rarediseases.org>

HSE Childhood Disability: <https://www2.hse.ie/services/disability-support-and-services-if-you-have-just-found-out-your-child-has-a-disability/useful-childhood-disability-websites.html>

Common congenital anomalies detected in pregnancy

Nervous system anomalies: Neural Tube Defects

<https://www.cdc.gov/ncbddd/birthdefects/facts-about-neural-tube-defects.html>

Eye anomalies: Anophthalmos / Microphthalmos

<https://www.cdc.gov/ncbddd/birthdefects/anophthalmia-microphthalmia.html>

Congenital Heart Defects: sever CHD / Ventricular septal defect

<https://www.nhs.uk/conditions/congenital-heart-disease/> <https://www.cdc.gov/ncbddd/heartdefects/ventricularseptaldefect.html>

Respiratory anomalies: Congenital pulmonary airway malformations (CPAM)

<https://www.chop.edu/conditions-diseases/congenital-cystic-adenomatoid-malformation-ccam>

Oro-facial clefts: Cleft lip with or without cleft palate

<http://cleftcareireland.ie/cleft-lip-and-palate-explained/what-effects-can-a-cleft-have-on-my-child> <https://www.cdc.gov/ncbddd/birthdefects/cleftlip.html>

Gastro-intestinal anomalies: Diaphragmatic hernia

<https://www.cdc.gov/ncbddd/birthdefects/diaphragmatichernia.html>

Abdominal wall defects: Omphalocele

<https://www.cdc.gov/ncbddd/birthdefects/omphalocele.html>

Congenital anomalies of kidney and urinary tract (CAKUT):

Congenital hydronephrosis including ureter obstruction

<https://www.nhs.uk/conditions/hydronephrosis/>

Genital anomalies: Hypospadias

<https://www.cdc.gov/ncbddd/birthdefects/hypospadias.html>

Limb anomalies: Club foot – talipes equinovarus

<https://www2.hse.ie/conditions/positional-talipes/>

Other anomalies / syndromes: Vascular disruption anomalies

Genetic disorders: Trisomy 21

<https://www.cdc.gov/ncbddd/birthdefects/surveillancemanual/quick-reference-handbook/trisomy-21-down-syndrome.html>

Common congenital anomalies detected in a newborn

Nervous system anomalies: Neural Tube Defects

<https://www.cdc.gov/ncbddd/birthdefects/facts-about-neural-tube-defects.html>

<https://www.cdc.gov/ncbddd/birthdefects/surveillancemanual/quick-reference>

[-handbook/spinabifida.html](#)

Eye anomalies: Congenital Cataract

<https://www.nhs.uk/conditions/childhood-cataracts/>

Ear, Face and Neck Anomalies: Anotia and atresia / stenosis / stricture of external auditory canal

<https://www.cdc.gov/ncbddd/birthdefects/anotia-microtia.html>

Congenital Heart Defects: sever CHD / Ventricular septal defect

<https://www.nhs.uk/conditions/congenital-heart-disease/>

Respiratory anomalies: Congenital pulmonary airway malformations (CPAM)

<https://www.chop.edu/conditions-diseases/congenital-cystic-adenomatoid-malformation-ccam>

Oro-facial clefts: Cleft palate <http://cleftcareireland.ie>

Gastro-intestinal anomalies: Ano-rectal atresia and stenosis

<https://rarediseases.org/rare-diseases/imperforate-anus/>

Abdominal wall defects: Omphalocele

<https://www.cdc.gov/ncbddd/birthdefects/omphalocele.html>

Congenital anomalies of kidney and urinary tract (CAKUT): Congenital hydronephrosis including ureter obstruction

<https://www.nhs.uk/conditions/hydronephrosis/>

Genital anomalies: Hypospadias

<https://www.cdc.gov/ncbddd/birthdefects/hypospadias.html>

Limb anomalies: Hip Dislocation

<https://www.olhc.ie/services/departments-a-z/departments-of-paediatric-orthopaedic-surgery/conditions-we-treat/developmental-dysplasia-of-the-hip--ddh-/developmental-hip-dysplasia-a-guide-for-parents.pdf>

Other anomalies / syndromes:

VATER / VACTERL association

<https://rarediseases.org/rare-diseases/vacterl-association/>

Teratogenic syndromes resulting in major malformations eg congenital rubella infection

<https://www.childrenshospital.org/conditions/congenital-rubella>

Genetic disorders: Trisomy 21

<https://www.cdc.gov/ncbddd/birthdefects/surveillancemanual/quick-reference-handbook/trisomy-21-down-syndrome.html>

How to reduce the risk of birth defects.

Pre-pregnancy and pregnancy care: Taking a daily prenatal vitamin that includes at least 400 micrograms of folic acid can help prevent a variety of birth defects. In addition to taking your prenatal vitamin, avoiding alcohol, smoking, and illegal drugs can help prevent birth defects and pregnancy complications.

<https://www2.hse.ie/wellbeing/pregnancy-and-birth/trying-for-a-baby/food-and-supplements-for-a-healthy-pregnancy/>

<https://www2.hse.ie/wellbeing/pregnancy-and-birth/keeping-well/food-drink/>

Awareness of past or current conditions. If you previously had a pregnancy with a birth defect, it's important to find out the most likely causes because it can help your doctor plan preventive measures for your next pregnancy.

EPIDEMIOLOGY: CORK & KERRY CONGENITAL ANOMALY REGISTER 2020 DATA

There were a total of 7,914 live births (4,072 Males and 3,842 Females) in Cork & Kerry in 2020. There were 26 stillbirths in 2020. The number of babies born with a birth defect was 206 (3%), 15 twin deliveries, 191 singleton. Of these, there were 98 male, 97 female and 11 babies where the sex was not documented.

Mother's age at delivery where the child had a congenital anomaly in 2020 is shown in Table 1. The prevalence of delivering a baby with a congenital anomaly increases with maternal age from age 35+.

TABLE 1: BIRTHS BY MATERNAL AGE 2020 IN CORK & KERRY

Age Group	All Live Births 2020	Births with Congenital anomalies*	Rate/1000 Live births
15-19	89	<5	22.47
20-24	465	12	25.80
25-29	1186	26	21.92
30-34	2789	44	15.77
35-39	2675	90	33.64
40-44	660	32	48.48
45+	50	0	0
Total	7914	206	26.02

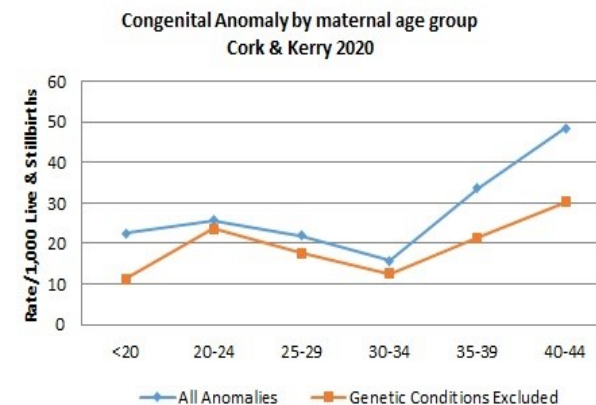
* 2020 Data extracted from Registry 09th December 2022

In 12 (46%) of the stillbirths registered in Cork & Kerry during 2020, a congenital anomaly was present. Stillbirths accounted for 0.3% of total births in Cork & Kerry in 2020. There were 28 terminations of pregnancy

There were 166 live births of babies with a congenital anomaly in the Cork & Kerry region in 2020. See Table 2 below.

TABLE 2: TYPE OF BIRTH 2020 CORK & KERRY

Type of Birth	Number of Cases	%
Live birth	166	80%
Stillbirth or fetal death >=20 weeks gestation	12	6%
Termination of Pregnancy	28	14%
Total number of infants	206	100%



The diagnosis of a congenital anomaly in a child can be identified at birth but in many instances it can be a delayed diagnosis. In 2020, 1 baby with Congenital Anomaly was discovered >12 months.

See Table 3 below when a congenital anomaly diagnosis was discovered in Cork and Kerry babies in 2019.

TABLE 3: WHEN DIAGNOSIS WAS DISCOVERED IN CORK & KERRY IN 2020

When Discovered	Cases	%
Prenatal diagnosis	92	45%
At birth	68	33%
Less than 1 week	20	10%
1-4 weeks	6	3%
1-12 months	19	9%
Over 12 months	<5	0%
Total	206	100%

In 2020, the gestational age range for babies born with a congenital anomaly ranged between 12 to 42 weeks. A baby born with a congenital anomaly is more likely to be born prematurely and to have a low birth weight. See Tables 4 and 5.

TABLE 4 GESTATIONAL AGE OF INFANTS ON CORK & KERRY REGISTER 2020

Length of Gestation 2020	No (%) of infants registered on Cork & Kerry Register 2020	No (%) of infants nationally 2020
Under 35	48 (23%)	2, 259 (4%)
36 & over	158 (77%)	54,364 (96%)
Not Stated	0 (0%)	189 (0%)
Total	206 (100%)	56,812 (100%)

– CSO Annual Report on Vital Statistics 2020.

^ Stillbirth data - Not all Stillbirths are registered. Registration of a stillbirth is voluntary. The numbers recorded by the CSO for Cork & Kerry in 2020 is 17. This is an underestimate of the true number of 26 for this region. As registration is voluntary, only the numbers of stillborn births that are registered are published. This number has been obtained by the Registry from the Birth Notification Form and the Labour Ward Registry.

TABLE 5 INFANT BIRTH WEIGHT CORK & KERRY REGISTER 2020

Birth weight 2020	No (%) Infants in Cork & Kerry Register 2020	No (%) of infants nationally 2020 ¹
Under 2499g	43 (21%)	3137 (6%)
2500g & over	163 (79%)	53552 (94%)
Not stated	0 (0%)	123 (0%)
Total	206 (100%)	56,812 (100%)

See tables 6 and 7 for prevalence and aetiology of congenital anomalies registered for 2020 in our region.

TABLE 6 CASES AND PREVALENCE PER 10,000 BIRTHS FROM CORK & KERRY REGISTRY DATA COMPARED TO EUROCAT FULL MEMBER REGISTRY DATA, 2020

(INCLUDES LIVE BIRTHS, GENETIC CONDITIONS, FETAL DEATHS, AND TERMINATIONS OF PREGNANCY FOR FETAL ANOMALY FOLLOWING PRENATAL DIAGNOSIS WHERE DATA IS AVAILABLE)

Congenital Anomaly	Cork & Kerry Cases	Cork & Kerry Prevalence RATE per 10,000 births	EUROCAT 2020 Prevalence RATE per 10,000 births https://eu-rd-platform.jrc.ec.europa.eu/eurocat/eurocat-data/prevalence_en
ALL ANOMALIES	206	262.79	253.38 (249.26 - 257.55)
Anomaly			
Nervous system	17	21.69	26.03 (24.72 - 27.39)
Eye	<5	5.1	3.51 (3.04 - 4.03)
Ear, face and neck	<5	5.1	1.51 (1.21 - 1.86)
Congenital heart defects	90	114.81	79.70 (77.40 - 82.06)
Respiratory	<5	3.83	3.91 (3.42 - 4.46)
Oro-facial clefts	9	11.48	14.38 (13.41 - 15.39)
Digestive system	15	19.14	17.27 (16.21 - 18.38)
Abdominal wall defects	<5	3.83	7.37 (6.68 - 8.11)
Urinary	19	24.24	35.54 (34.01 - 37.12)
Genital	13	16.58	18.69 (17.59 - 19.85)
Limb	50	63.78	37.17 (35.61 - 38.79)
Chromosomal	61	77.82	61.73 (59.71 - 63.80)

*NOTE: Table extracted for Cork & Kerry Register on Dec 12th and from the EUROCAT website data prevalence reports on accessed on 19/1/2023. The total number of cases on the Cork & Kerry Congenital Anomaly Register for the year 2020 are (n=206) .

TABLE 7 MULTIPLE MALFORMATION OF CONGENITAL ANOMALY 2020 CORK & KERRY

Multiple Malformations 2020	No. Cases	%
Isolated cardiac	50	24%
Genetic syndrome, skeletal dysplasia and monogenic disorder	10	5%
Chromosomal	51	25%
Isolated other	52	25%
Multiple anomalies	26	13%
NTD isolated	<5	2%
Isolated renal	12	6%
Teratogenic syndromes (CMV)	0	0%
Total	206	100%

Half of all anomalies are isolated (cardiac 24%; other isolated 25%). Multiple anomalies were present in 26 births (13%). There is a known chromosomal (25%) or other genetic cause (5%) in about 30% of cases. In 2020 there were no cases of birth defect secondary to maternal cytomegalovirus (CMV) infection during pregnancy.

A word of thanks to the staff of Cork & Kerry Hospitals who facilitate our Research Nurse in assisting with case ascertainment and accessing medical notes to update the registry.

Location of HSE Congenital Anomaly Registries in Ireland



For further information on Congenital Anomaly Registers in Ireland go to our website below at



<http://www.hse.ie/congenitalanomalyregistersireland>

<http://www.eurocat-network.eu/>