Public Health England

Protecting and improving the nation's health

# National Congential Anomaly and Rare Disease Registration Service

## Congenital anomaly statistics 2018: technical details

# About Public Health England

Public Health England exists to protect and improve the nation's health and wellbeing, and reduce health inequalities. We do this through world-leading science, research, knowledge and intelligence, advocacy, partnerships and the delivery of specialist public health services. We are an executive agency of the Department of Health and Social Care, and a distinct delivery organisation with operational autonomy. We provide government, local government, the NHS, Parliament, industry and the public with evidence-based professional, scientific and delivery expertise and support

Public Health England
Wellington House
133-155 Waterloo Road
London SE1 8UG
Tel: 020 7654 8000
www.gov.uk/phe
Twitter: @PHE_uk
Facebook: www.facebook.com/PublicHealthEngland



Published August 2020
PHE publications
gateway number: GW-1445

PHE supports the UN
Sustainable Development Goals

# Contents

## Incidence and birth prevalence

Incidence is the total number of 'new' cases of disease occurring in a population in a specified time period, whereas prevalence is the total number of 'all' cases in a population at one point in time. Conventionally, as in this report, congenital anomaly registers report prevalence estimates. This is because it is not possible to ascertain all 'new' babies with any particular anomaly, as a proportion of pregnancies affected with an anomaly will miscarry spontaneously before being diagnosed. There is not a population estimate of the total number of pregnancies at risk of being affected by an anomaly due to miscarriages and terminations of pregnancy for fetal anomaly (TOPFA). Therefore, congenital anomaly registers report prevalence estimates per 1,000 or 10,000 total births (live and stillbirths). By convention, these are referred to as birth prevalence estimates even though the pregnancy may not result in a registered birth because of late miscarriage or TOPFA.

## Confidence intervals

Confidence intervals are calculated around many different types of statistic used in public health analysis. Comparisons are often made between 2 or more different rates. In this report, examples include estimated birth prevalence comparisons between different regions of registration, age groups and type of congenital anomaly. Statistical testing is undertaken by comparing the confidence intervals of estimated birth prevalence to see if they overlap - with non-overlapping confidence intervals being considered as statistically significantly different.

The confidence intervals used in this report are calculated using the Poisson distribution.[1] More information about the use of confidence intervals and the reasons for using them are explained in APHO Technical Briefing 3 – Commonly used public health statistics and their confidence intervals.[1]

## Calculation of birth prevalence and their 95% confidence intervals

$$\text{Birth prevalence} = \frac{\text{Number of cases (live births + stillbirths + late miscarriages + TOPFAs)}}{\text{Number of births (live births+stillbirths)}} \times 10{,}000$$

$$\text{Lower 95\% confidence limit} = \frac{\left(\frac{1.96}{2} - \sqrt{\text{number of cases} + 0.02}\right)^2}{\text{number of births}} \times 10{,}000$$

$$\text{Upper 95\% confidence limit} = \frac{\left(\frac{1.96}{2} + \sqrt{\text{number of cases} + 0.96}\right)^2}{\text{number of births}} \times 10{,}000$$

[1] Bégaud B, Martin K, Abouelfath A, Tubert-Bitter P, Moore N, Moride Y. An easy to use method to approximate

## Data collection

A congenital anomaly is defined as any defect present at delivery, probably originating before birth, and includes structural, chromosomal and other genetic anomalies. Some congenital anomalies are detected during pregnancy, some are found at birth, while others become obvious only as a baby grows older.

Congenital anomaly data are collected from a number of different sources including:

- maternity units
- neonatal units
- diagnostic departments (paediatric, neonatal, clinical genetics, antenatal ultrasound, fetal medicine, pathology)
- genetic laboratories
- NHS Trust IT departments
- child health systems
- local audit schemes
- disease-specific registers
- neighbouring national registers

This multiple source reporting enables NCARDRS to achieve the highest possible ascertainment of congenital anomalies in the population. Much of the focus to date has been on ensuring high ascertainment and completeness of cases and ensuring consistency and standardisation across the country.

A single data management system has been developed and NCARDRS has a team of around 45 dedicated registration officers and analysts. NCARDRS currently takes electronic data from over 500 NHS providers across the country.

Data is collected on all suspected and confirmed congenital anomalies identified in utero, at birth or at any point in childhood. In addition to babies that are liveborn or stillborn that have congenital anomalies, information about terminations of pregnancy with a diagnosed fetal anomaly at any gestation (TOPFA) and miscarriages where an anomaly is present, is also collected. NCARDRS only report on late miscarriages (20 to 23 weeks' gestation) as ascertainment of all miscarriages with congenital anomalies is not possible.

NCARDRS collects information about the mother and child, including postcode of residence, mother's age, pregnancy length, pregnancy outcome, when and how the anomaly was identified and the details of each anomaly. Some identifiable information is collected on the mother and child but only enough information to avoid duplicate

---

Poisson confidence limits. *European Journal of Epidemiology* 2005; 20:213-216.

registrations and for the validations of cases, ensuring accurate matching between antenatally diagnosed anomalies and postnatal notifications.

## Data quality

All 10 reporting regions submitted data to the European Surveillance of Congenital Anomalies (EUROCAT) and followed their data quality procedures, ensuring collection of a number of core variables. More information can be found in the Guidelines for data registration section of the EUROCAT website. In addition, there is an established national process and system for data collection, processing and quality assurance, adopting internationally approved methods of coding, recording, and analysis.

## Inclusion criteria

All livebirths, fetal deaths with gestational age (GA) ≥20 weeks and TOPFA (at any gestational age) with at last one registered anomaly are included for reporting.

## Reporting

NCARDRS uses the EUROCAT congenital anomaly subgroup categories for reporting (see tables below). These subgroups use ICD10 codes to group together conditions by body system and anomaly type.

## EUROCAT congenital anomaly subgroups

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| **All anomalies** | Q-chapter, D215, D821, D1810, P350, P351, P371 | | Exclude all minor anomalies as specified in exclusion list below |
| **Nervous system** | Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07 | | Q0461, Q0782 |
| Neural tube defects | Q00, Q01, Q05 | | |
| Anencephalus and similar | Q00 | | |
| Encephalocele | Q01 | Exclude if associated with anencephalus subgroup | |

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| Spina Bifida | Q05 | Exclude if associated with anencephalus or encephalocele subgroups | |
| Hydrocephalus | Q03 | Exclude hydranencephaly. Exclude association with NTD subgroup | |
| Severe Microcephaly | Q02 | Exclude association with NTD subgroup | |
| Arhinencephaly / holoprosencephaly | Q041, Q042 | | |
| **Eye** | Q10-Q15 | | Q101-Q103, Q105, Q135 |
| Anophthalmos / microphthalmos | Q110, Q111, Q112 | | |
| Anophthalmos | Q110, Q111 | | |
| Congenital cataract | Q120 | | |
| Congenital glaucoma | Q150 | | |
| **Ear, face and neck** | Q16, Q17, Q18 | | Q170-Q175, Q179, Q180-Q182, Q184-Q187, Q1880, Q189 |
| Anotia | Q160 | | |
| **Congenital Heart Defects** | Q20-Q26 | Exclude PDA with GA <37 weeks Exclude peripheral pulmonary artery stenosis with GA < 37 weeks | Q2111, Q250 if GA <37 weeks, Q2541, Q256 if GA<37 weeks, Q261 |

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| Severe CHD | Q200, Q201, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q232, Q233, Q234, Q251, Q252, Q262 | | |
| Common arterial truncus | Q200 | | |
| Double outlet right ventricle | Q201 | | |
| Transposition of great vessels | Q203 | | |
| Single ventricle | Q204 | | |
| Ventricular septal defect | Q210 | | |
| Atrial septal defect | Q211 | | Q2111 |
| Atrioventricular septal defect | Q212 | | |
| Tetralogy of Fallot | Q213 | | |
| Tricuspid atresia and stenosis | Q224 | | |
| Ebstein's anomaly | Q225 | | |
| Pulmonary valve stenosis | Q221 | | |
| Pulmonary valve atresia | Q220 | | |
| Aortic valve atresia/stenosis | Q230 | | |
| Mitral valve anomalies | Q232, Q233 | | |
| Hypoplastic left heart | Q234 | | |
| Hypoplastic right heart | Q226 | | |
| Coarctation of aorta | Q251 | | |
| Aortic atresia / interrupted aortic arch | Q252 | | |
| Total anomalous pulm venous return | Q262 | | |
| Patent ductus arteriosus as **only** CHD in term infants (GA +37 weeks) | Q250 | Livebirths only | |
| **Respiratory** | Q300, Q32-Q34 | Exclude Q336 | Q320, Q331 |
| Choanal atresia | Q300 | | |
| Cystic adenomatous malformation of lung | Q3380 | | |
| **Oro-facial clefts** | Q35-Q37 | Exclude association with holoprosencephaly or anencephaly subgroups | |

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| Cleft lip with or without cleft palate | Q36, Q37 | Exclude association with holoprosencephaly or anencephaly subgroups | |
| Cleft palate | Q35 | Exclude association with cleft lip subgroup. Exclude association with holoprosencephaly or anencephaly subgroups | |
| **Digestive system** | Q38-Q45, Q790 | | Q381, Q382, Q3850, Q400, Q401. Q4021, Q430, Q4320, Q4381, Q4382 |
| Oesophageal atresia with or without tracheo-oesophageal fistula | Q390-Q391 | | |
| Duodenal atresia or stenosis | Q410 | Exclude if also annular pancreas subgroup | |
| Atresia or stenosis of other parts of small intestine | Q411-Q418 | | |
| Ano-rectal atresia and stenosis | Q420-Q423 | | |
| Hirschsprung's disease | Q431 | | |
| Atresia of bile ducts | Q442 | | |
| Annular pancreas | Q451 | | |
| Diaphragmatic hernia | Q790 | | |
| **Abdominal wall defects** | Q792, Q793, Q795 | | |
| Gastroschisis | Q793 | | |
| Omphalocele | Q792 | | |
| **Urinary** | Q60-Q64, Q794 | | Q610, Q627, Q633 |
| Bilateral renal agenesis including Potter syndrome | Q601, Q606 | Exclude unilateral | |
| Multicystic Renal dysplasia | Q6140, Q6141 | | |

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| Congenital hydronephrosis | Q620 | | |
| Bladder exstrophy and / or epispadia | Q640, Q641 | | |
| Posterior urethral valve and / or prune belly | Q6420, Q794 | | |
| **Genital** | Q50-Q52, Q54-Q56 | | Q523, Q525, Q527, Q5520, Q5521 |
| Hypospadias | Q54 | | |
| Indeterminate sex | Q56 | | |
| **Limb** | Q65-Q74 | | Q653-Q656, Q662-Q669, Q670-Q678, Q680, Q6810, Q6821, Q683-Q685, Q7400 |
| Limb reduction | Q71-Q73 | | |
| Club foot – talipes equinovarus | Q660 | | |
| Hip dislocation and / or dysplasia | Q650–Q652, Q6580, Q6581 | | |
| Polydactyly | Q69 | | |
| Syndactyly | Q70 | | |
| **Other anomalies / syndromes** | | | |
| Skeletal dysplasias | Q7402, Q77, Q7800, Q782-Q788 | | |
| Craniosynostosis | Q750 | | |
| Congenital constriction bands / amniotic band | Q7980 | | |
| Situs inversus | Q893 | | |
| Conjoined twins | Q894 | | |
| Congenital skin disorders | Q80-Q82 | | Q825, Q8280 |
| VATER/VACTERL | Q8726 | | |

| Subgroups | ICD10-BPA | Comments | Excluded minor anomalies |
|---|---|---|---|
| Vascular disruption anomalies | Q0435, Q411, Q412, Q418, Q710, Q712, Q713, Q720, Q722, Q723, Q730, Q793, Q795, Q7980, Q7982, Q8706 | | |
| Laterality anomalies | Q206, Q240, Q3381, Q890, Q893 | | |
| Teratogenic syndromes with malformations | Q86, P350, P351, P371 | | |
| Fetal alcohol syndrome | Q860 | | |
| Valproate syndrome | Q8680 | | |
| Maternal infections resulting in malformations | P350, P351, P371 | | |
| Genetic syndromes + microdeletions | Q4471, Q6190, Q7484, Q751, Q754, Q7581, Q87, Q936, D821 | Exclude Associations and sequences Exclude Q8703,Q8704, Q8706,Q8708, Q8724,Q8726 | |
| **Chromosomal** | Q90-Q92, Q93, Q96-Q99 | Exclude microdeletions Q936 | |
| Down's syndrome | Q90 | | |
| Patau's syndrome | Q914-Q917 | | |
| Edwards' syndrome | Q910-Q913 | | |
| Turner's syndrome | Q96 | | |
| Klinefelter's syndrome | Q980-Q984 | | |

## List of exclusions

| | Specified ICD10-BPA – if present |
|---|---|
| **Head** | |
| Aberrant scalp hair patterning | |
| Brachycephaly | |
| Flat occiput | |
| Depressions in skull, lacunar skull, temporal flattening | Q6740 |
| Dolichocephaly | Q672 |
| Plagiocephaly – head asymmetry | Q673 |
| Third fontanelle | |
| Macrocephalus | Q753 |
| Facial asymmetry | Q670 |
| Compression facies | Q671 |
| Other cong deformities of skull, face and jaw (including all types of abnormally shaped skull without synostosis) | Q674 |
| Skull, late closure | |
| Dysmorphic face | Q189 |
| Broad, prominent forehead | |
| Coarse facies | |
| Flattened face | |
| Frontal bossing / wide forehead | |
| Mid face hypoplasia | |
| Pointed facies | |
| Round head shape | |
| Sloping forehead | |
| Metopic ridge, high metopic suture | |
| Wormian bones | |
| Bony occipital spur | |
| **Eyes** | |
| Anisocoria | |
| Dacryocystocele | H046 |
| Epicanthic folds | Q189 |
| Epicenthus inversus | Q189 |
| Exophthalmos | H052 |
| Upward slanting palpebral fissures | Q103 |
| Downward slanting palpebral fissures | Q103 |
| Short palpebral fissures | Q189 |
| Congenital ectropion | Q101 |
| Congenital entropion | Q102 |
| Other congenital malformations of eyelid | Q103 |
| Oval shaped pupils | |
| Prominent/protruding eyes | H052 |
| Dystopia canthorum | Q189 |
| Hypertelorism | Q752 |
| Hypotelorism | Q189 |

| | Specified ICD10-BPA – if present |
|---|---|
| Stenosis of stricture of lacrimal duct | Q105 |
| Synophrys | Q1880 |
| Blue sclera | Q135 |
| Crocodile tears | Q0782 |
| **Ears** | |
| Primitive shape | Q173 |
| Lack of helical fold | Q173 |
| Asymmetric size | Q173 |
| Posterior angulation | Q173 |
| Microtia/small ears | Q172 |
| Macrotia | Q171 |
| Protuberant ears | Q173 |
| Absent tragus | |
| Double lobule | Q170 |
| Accesorry auricle, preauricular appendage, tag or lobule | Q170 |
| Auricular pit | |
| Pointed ear, Vulcan ear, simple ear | Q173 |
| Preauricular sinus or cyst | Q181 |
| Narrow external auditory meatus | |
| Low set ears | Q174 |
| Bat ear, prominent proturberant ear | Q175 |
| Congenital absence of ear lobe | |
| Darwin's tubercle | |
| Unspecified and minor malformation of ear | Q179 |
| **Nose** | |
| Small/hypoplastic nares | Q189 |
| Notched alas | |
| Anteverted nares | Q189 |
| Bifid tip of nose | Q189 |
| Broad nasal root, anomaly of nasal root | Q189 |
| Depressed nasal bridge | Q189 |
| Deviation of nasal septum | Q6741 |
| Dysmorphic nose | Q189 |
| Flat nose | Q189 |
| Flattened nasal bridge | Q189 |
| Pinched nose | Q189 |
| Prominent nasal bridge | Q189 |
| Saddle nose | Q189 |
| Small pointed nose | Q189 |
| Underdeveloped nasal bones | Q189 |
| Upturned nose | Q189 |
| Wide nasal root | Q189 |
| **Oral regions** | |
| Borderline small mandible/ minor micrognathia | |
| Aberrant frenula | |
| Absent /hypoplasia depressor anguli oris (asymmetric crying face) | |

| | Specified ICD10-BPA – if present |
|---|---|
| Alveolar crest | |
| Anomalies of philtrum, elongated philtrum | Q189 |
| Bifid uvula / cleft uvula | Q357 |
| Borderline small mandible/ minor micrognathia | |
| Disturbances in tooth eruption | |
| Enamel hypoplasia | |
| Glossoptosis | |
| Malformed teeth | |
| High arched palate | Q3850 |
| Tongue tie or cyst of tongue | Q381 |
| Macroglossia / hemi-hypertrophy of tongue | Q382 |
| Macrostomia | Q184 |
| Malformed teeth | |
| Microstomia | Q185 |
| Macrocheilia | Q186 |
| Microcheilia | Q187 |
| Microglossia | |
| Microstomia | Q189 |
| Mid-oral tongue position | |
| Neonatal teeth | |
| Prominent jaw | Q189 |
| Retrognathia/ receding chin | Q674 |
| Short philtrum | Q189 |
| Thin lips | Q189 |
| Ranula | |
| **Neck** | |
| Broad neck | Q189 |
| Congenital thymic hypoplasia | |
| Short neck | Q189 |
| Mild webbed neck | |
| Sinus, fistula or cyst of branchial cleft | Q180 |
| Thymus involution | |
| Thyreoglossal cyst | |
| Preauricular sinus or cyst | Q181 |
| Other branchial cleft malformations | Q182 |
| Congenital malformation of face and neck, unspecified | Q189 |
| Torticollis | Q680 |
| **Hands** | |
| Duplication of thumbnail | |
| Arachnodactyly | |
| Enlarged or hypertrophic nails | Q845 |
| Single/abnormal palmar crease | Q8280 |
| Unusual dermatoglyphics | |
| Clinodactyly (5th finger) | Q6810 |
| Short fingers (4th. 5th finger) | |
| Accessorry carpal bones | Q7400 |

| | Specified ICD10-BPA – if present |
|---|---|
| Other congenital malformations of nails | Q846 |
| Overlapping fingers | |
| Small fingers | |
| Subluxation of phalangeal bones | |
| **Feet -Limb** | |
| Bulbous toes | |
| Syndactyly (2nd to 3rd toes) | |
| Gap between toes (1st to 2nd) | |
| Short great toe | |
| Recessed toes (4th, 5th) | |
| Enlarged or hypertrophic nails | Q845 |
| Prominent calcaneus | |
| Clicking hip, subluxation of unstable hip | Q653-Q65.6 |
| Metatarsus varus or metatarsus adductus | Q662 |
| Hallux varus – other cong varus deformities of feet | Q663 |
| Talipes or pes calcaneovalgus | Q664 |
| Talipes calcaneovarus | Q661 |
| Congenital pes planus | Q665 |
| Hip dysplasia and other specified/unspecified hip anomalies | Q658, Q659 |
| Metatarsus varus – other cong valgus deformities of feet | Q666 |
| Overlapping toes | |
| Pes cavus | Q667 |
| Rocker bottom feet | Q6680 |
| Clubfoot of postural origin – other cong deformities of feet | Q668 |
| Congenital deformity of feet, unspecified | Q669 |
| **Skin** | |
| Hemangioma if no treatment is required | |
| Pigmented naevus – cong non-neoplastic naevus | Q825 |
| Neavus flammeus | Q8250 |
| Strawberry naevus | Q8251 |
| Lymphangioma if no treatment is required | |
| Angioma | |
| Persistent lanugo | |
| Mongoloid spot (whites) | Q8252 |
| Depigmented spot | |
| Unusual placement of nipples/wide spaced nipples | |
| Accessory nipples | Q833 |
| Accessory skin tags | Q8281 |
| Cafe-au-lait spot | |
| Epibulbar dermoid | |
| Heterochromia of hair | |
| Hypoplasia of toe nails | Q846 |
| **Skeletal** | |
| Cubitus valgus | |
| Prominent sternum / pectus carinatum | Q677 |
| Prominent sternum | |

| | Specified ICD10-BPA – if present |
|---|---|
| Depressed sternum / pectus excavatum | Q676 |
| Sternum bifidum | Q7671 |
| Shieldlike chest, other cong deformities of chest | Q678 |
| Congenital deformity of spine | Q675 |
| Genua valgum | |
| Genus varum | |
| Genu recurvatum | Q6821 |
| Duplication of ribs | |
| Congenital bowing of femur | Q683 |
| Congenital bowing of fibula and tibia | Q684 |
| Congenital bowing of long bones of leg, unspecified | Q685 |
| Congenital bowing of upper limb | |
| No ossification of os coccyx | |
| Ovoid configuration of vertebrae | |
| Spina bifida occulta | Q760 |
| Sacral dimple | L059 |
| Cervical rib | Q765 |
| Fused rib, single | |
| Absence of rib/hypoplastic rib | Q7660 |
| Accessory rib | Q7662 |
| Congenital lordosis, postural | Q7643 |
| Abortive 12th rib | |
| Coronal clefts of vertebrae, incomplete | |
| Bipartite vertebrae | |
| Bifid ribs | |
| **Brain** | |
| Arachnoid cysts | |
| Asymmetric ventricles, normal size | |
| Banana shaped cerebellum | |
| Cerebellar hypoplasia, mild | |
| Cerebral atrophy | |
| Cyst of septum pellucidum | |
| Enlarged cisterna magna, isolated | |
| Jaw-winking syndrome, Marcus Gunn's syndrome | Q0780 |
| Periventricular leukomalacia | |
| Single congenital cerebral cyst | Q0461 |
| Thin or hypoplastic corpus callosum | |
| Ventriculomegaly < 15 mm | |
| Choroid plexus cysts | |
| Anomalies of septum pellucidum | |
| **Cardiovascular** | |
| Absence or hypoplasia of umbilical artery, single umbilical artery | Q270 |
| Absence of vena cava superior | |
| Functional or unspecified cardiac murmur | R011 |
| Cardiomegaly | I517 |
| Cardiomyopathy | I429 |

| | Specified ICD10-BPA – if present |
|---|---|
| Deviation of the heart axis | |
| Patent ductus arteriosus if GA < 37 weeks | Q250 if GA <37 weeks |
| Peripheral pulmonary artery stenosis | Q256 if GA<37 weeks |
| Patent or persistent foramen ovale | Q2111 |
| Persistent left superior vena cava | Q261 |
| Persistent right aortic arch | Q2541 |
| Persistent right umbilical vein | |
| Congenital heart block | Q246 |
| **Pulmonary** | |
| Accessory lobe of lung | Q331 |
| Congenital laryngeal stridor | Q314 |
| Laryngomalacia | Q3140 |
| Tracheomalacia | Q320 |
| Azygos lobe of lung | Q3310 |
| Bronchomalacia | Q322 |
| Single cyst of the lung | Q3300 |
| Hyperplasia of thymus | |
| Pleural effusion | |
| Pulmonary hypoplasia, secondary | |
| Relaxation of diaphragm | |
| Thymus involution | |
| Vocal cord palsy | |
| **Gastro-intestinal** | |
| Hiatus hernia | Q401 |
| Abdominal cyst not needing surgery | |
| Accessory spleen | |
| Choledochal cyst | Q444 |
| Congenital adrenal hypoplasia | Q8911 |
| Congenital cholestasis | |
| Congenital mesenteric cyst | Q4583 |
| Cyst of spleen | |
| Dilatation of intestine | |
| Hepatomegaly | R160 |
| Liver cyst | |
| Plica of anus | |
| Splenomegaly | |
| Pyloric stenosis | Q400 |
| Diastasis recti | |
| Umbilical hernia | |
| Inguinal hernia | K409 |
| Meckel’s diverticulum | Q430 |
| Functional gastro-intestinal disorders | Q4021, Q4320, Q4381, Q4382 |
| Transient choledochal cyst | |
| Anterior anus without surgery | |

| | Specified ICD10-BPA – if present |
|---|---|
| **Renal** | |
| Vesico-ureteral-renal reflux | Q627 |
| Enlarged/thickened bladder | |
| Hydronephrosis with a pelvis dilatation less than 10 mm | |
| Hyperplastic and giant kidney | Q633 |
| Single renal cyst | Q610 |
| **External genitals** | |
| Deficient or hooded foreskin | |
| Undescended testicle | Q53 |
| Unspecified ectopic testis | |
| Retractile testis | Q5520 |
| Hydrocele of testis | |
| Phymosis | |
| Bifid scrotum | Q5521 |
| Buried penis | |
| Congenital chordee | Q544 |
| Congenital adrenogenital disorders | E250 |
| Congenital malformation of vulva | Q527 |
| Congenital torsion of ovary | Q502 |
| Curvature of penis | |
| Hypoplasia of penis / micropenis | |
| Hymen imperforate | Q523 |
| Fusion of labia | Q525 |
| Prominent labia minora | |
| Enlarged clitoris | |
| Vaginal skin tag | |
| Cysts of vulva | |
| Transient ovarian cyst | |
| Developmental ovarian cyst(s) | Q501, Q5010, Q5011 |
| Embryonic cyst of broad ligament | Q505 |
| Foreskin tethered to the scrotum | N47 |
| Hypertrophy of hymen | |
| Phimosis | N47 |
| Seminal vesicle cyst | |
| Testicular torsion | N44 |
| **Other** | |
| Congenital malformation, unspecified | Q899 |
| **Chromosomal** | |
| Balanced chromosomal rearrangements | Q95 |
| Balanced translocations or inversions in normal individuals | |
| Balanced autosomal rearrangement in abnormal individual | Q952 |
| Individuals with marker heterochromatin | |
| Individuals with autosomal fragile site | |

**"Non-congenital" anomalies**

Pyloric stenosis – there is controversy about the congenital nature of the majority of cases.

Patent ductus arteriosus in babies <37 weeks

Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births should be thoroughly checked before registration.

**Poorly specified anomalies**
Functional or unspecified cardiac murmur
Laryngomalacia and tracheomalacia
Functional gastro-intestinal disorders
Undescended testicle. Registries may choose to record this locally if they can follow-up all babies to ascertain whether the testis descends normally.
Unspecified ectopic testis
Vesico-ureteral reflux. Registries should record and transmit to EUROCAT the underlying anomaly, if present.
Clicking hip
Clubfoot where this is no further specification of whether malformation or postural origin

## Down’s syndrome, Edwards’ syndrome and Patau’s syndrome source data and completeness

Diagnostic: Data in electronic form are regularly sent to NCARDRS by every NHS cytogenetic laboratory in England, giving complete national ascertainment from this data feed. Laboratories follow a specific case definition, to ensure national consistency and data quality. Data are supplied for antenatal and postnatal testing (the latter category including fetal losses as well as livebirths), and for all test methods used in cytogenetics laboratories.

Outcome: Clinical outcome data are requested by regional NCARDRS teams from a variety of sources, including booking and delivery hospital, summary care record, antenatal screening teams and fetal medicine units. These data sources are less complete than the laboratory data for newer reporting regions of the country, however they are robust for regions that had previously established congenital anomaly registries.

## Down’s syndrome, Edwards’ syndrome and Patau’s inclusion and exclusion criteria

All babies with Down’s syndrome, Edwards’ syndrome or Patau’s syndrome delivered in 2018 with a confirmed cytogenetic laboratory diagnosis provided as part of care from NHS and private providers who submit data to NCARDRS are included within this report. This includes results obtained from conventional karyotyping (full or targeted), rapid aneuploidy testing (usually by FISH or QF-PCR), or microarray analysis. All specimen types are included, including prenatal (amniocentesis, chorionic villus sampling, fetal blood), postnatal (blood, buccal swab) and postmortem (solid tissue). When there is positive non-invasive prenatal testing (NIPT) and a clinical suspicion of Down’s syndrome, Edwards’ syndrome or Patau’s syndrome based on postnatal phenotype then these baby’s are also included in the figures.

## Geographical coverage of the NCARDRS regions in this report

| NCARDRS region | Local Authorities | |
|---|---|---|
| East Midlands and South Yorkshire | Amber Valley | Leicester |
| | Ashfield | Lincoln |
| | Barnsley | Mansfield |
| | Bassetlaw | Melton |
| | Balby | Newark and Sherwood |
| | Bolsover | North East Derbyshire |
| | Boston | North East Lincolnshire |
| | Broxtowe | North Kesteven |
| | Charnwood | North Lincolnshire |
| | Chesterfield | North West Leicestershire |
| | Corby | Northampton |
| | Daventry | Nottingham |
| | Derby | Oadby and Wigston |
| | Derbyshire Dales | Rotherham |
| | Doncaster | Rushcliffe |
| | East Lindsey | Rutland |
| | East Northamptonshire | Sheffield |
| | Erewash | South Derbyshire |
| | Gedling | South Holland |
| | Harborough | South Kesteven |
| | High Peak | South Northamptonshire |
| | Hinckley and Bosworth | Wellingborough |
| | Kettering | West Lindsey |
| Northern | Allerdale | Middlesbrough |
| | Carlisle | Newcastle upon Tyne |
| | Copeland | North Tyneside |
| | County Durham | Northumberland |
| | Darlington | Redcar and Cleveland |
| | Eden | South Tyneside |
| | Gateshead | Stockton-On-Tees |
| | Hartlepool | Sunderland |
| South West | Bath and North East Somerset | Plymouth |
| | Bristol, City of | Sedgemoor |
| | Cheltenham | South Gloucestershire |
| | Cornwall | South Hams |
| | Cotswold | South Somerset |
| | East Devon | Stroud |
| | Exeter | Swindon |
| | Forest of Dean | Taunton Deane Teignbridge |
| | Gloucester | Tewkesbury |
| | Isles of Scilly | Torbay |
| | Mendip | Torridge |
| | Mid Devon | West Devon |
| | North Devon | West Somerset |
| | North Somerset | Wiltshire (excluding Salisbury) |

| | | |
|---|---|---|
| Thames Valley | Aylesbury Vale | South Bucks |
| | Bracknell Forest | South Oxfordshire |
| | Cherwell | Vale of White Horse |
| | Chiltern | West Berkshire |
| | Milton Keynes | Windsor & Maidenhead |
| | Oxford | Wokingham |
| | Reading | Wycombe |
| | Slough | |
| Wessex | Basingstoke and Deane | North Dorset |
| | Bournemouth | Poole |
| | Christchurch | Portsmouth |
| | East Dorset | Purbeck |
| | East Hampshire | Rushmoor |
| | Eastleigh | Southampton |
| | Fareham | Test Valley |
| | Gosport | West Dorset |
| | Hart | Weymouth and Portland |
| | Havant | Wiltshire (Salisbury only) |
| | Isle of Wight | Winchester |
| | New Forest | |
| West Midlands | Birmingham | Shropshire |
| | Bromsgrove | Solihull |
| | Cannock Chase | South Staffordshire |
| | Coventry | Stafford |
| | Dudley | Stoke-on-Trent |
| | East Staffordshire | Stratford-on-Avon |
| | Herefordshire, County of | Tamworth |
| | Lichfield | Telford and Wrekin |
| | Malvern Hills | Walsall |
| | Newcastle-under-Lyme | Warwick |
| | North Warwickshire | Wolverhampton |
| | Nuneaton and Bedworth | Worcester |
| | Redditch | Wychavon |
| | Rugby | Wyre Forest |
| | Sandwell | |
| Yorkshire and Humber | Bradford | Leeds |
| | Calderdale | Richmondshire Ryedale |
| | Craven | Scarborough |
| | East Riding of Yorkshire | Selby |
| | Hambleton | Wakefield |
| | Harrogate | York |
| | Kingston upon Hull, City of | |
| | Kirkless | |