Public Health England

Protecting and improving the nation's health

National Congenital Anomaly and Rare Disease Registration Service

Congenital anomaly statistics 2019: technical details

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 a proportion of pregnancies affected with an anomaly will miscarry spontaneously before being identified. It is also not possible to provide a population estimate of the total number of pregnancies at risk of a congenital anomaly as there is no reliable estimate of the total number of pregnancies, given some result in miscarriage and terminations of pregnancy for fetal anomaly (TOPFA). Prevalence estimates are reported per 10,000 total births (live and stillbirths); 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, maternal age category and type of congenital anomaly. Basic 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 (Bégaud et al, 2005). More information about the use of confidence intervals is explained in the PHE Technical Guide.

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

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

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

Denominators

The number of total (live and still) births in England reported by the Office of National Statistics is used to calculate total birth prevalence and live birth prevalence of congenital anomalies. These are obtained from the PHE Data Lake. We also obtain the number of births from each of the UK Crown Dependencies; Isle of Man, Jersey and Guernsey. These are added to the total

births for the respective regions; North West and Wessex and also to the overall total births for England.

Geographical coverage of the NCARDRS regions

NCARDRS is made up of 10 reporting regions in England (Figure 1, main report). Regional boundaries used in this report are the EUROCAT reporting regions for England. To preserve the longitudinal trend, they are consistent with the legacy registers for these regions (East Midlands and South Yorkshire, Northern, South West, Thames Valley and Wessex). Table 1 lists the Local Authorities that are included in each region.

Table 1. Geographical coverage of the NCARDRS regions in this report

NCARDRS region	Local authorities	
East of England	Peterborough Luton Southend-on-Sea Thurrock Bedford Central Bedfordshire Cambridge East Cambridgeshire Fenland Huntingdonshire South Cambridgeshire Basildon Braintree Brentwood Castle Point Chelmsford Colchester Epping Forest Harlow Maldon Rochford Tendring Uttlesford	Broxbourne Dacorum Hertsmere North Hertfordshire Three Rivers Watford Breckland Broadland Great Yarmouth King's Lynn and West Norfolk North Norfolk Norwich South Norfolk Babergh Ipswich Mid Suffolk St Albans Welwyn Hatfield East Hertfordshire Stevenage West Suffolk East Suffolk

NCARDRS region	Local authorities	
East Midlands and South Yorkshire (EMSY)	Amber Valley Ashfield Barnsley Bassetlaw Balby Bolsover Boston Broxtowe Charnwood Chesterfield Corby Daventry Derby Derbyshire Dales Doncaster East Lindsey East Northamptonshire Erewash Gedling Harborough High Peak Hinckley and Bosworth Kettering	Leicester Lincoln Mansfield Melton Newark and Sherwood North East Derbyshire North East Lincolnshire North Kesteven North Lincolnshire North West Leicestershire Northampton Nottingham Oadby and Wigston Rotherham Rushcliffe Rutland Sheffield South Derbyshire South Holland South Kesteven South Northamptonshire Wellingborough West Lindsey

NCARDRS region	Local authorities	
London and South East	Medway Brighton and Hove Eastbourne Hastings Lewes Rother Wealden Ashford Canterbury Dartford Dover Gravesham Maidstone Sevenoaks Shepway Swale Thanet Tonbridge and Malling Tunbridge Wells Elmbridge Epsom and Ewell Guildford Mole Valley Reigate and Banstead Runnymeade Spelthorne Surrey Heath Tandridge Waverley Woking Adur Arun Chichester Crawley Horsham	Mid Sussex Worthing City of London Barking and Dagenham Barnet Bexley Brent Bromley Camden Croyden Ealing Enfield Greenwich Hackney Hammersmith and Fulham Haringey Harrow Havering Hillingdon Hounslow Islington Kensington and Chelsea Kingston upon Thames Lambeth Lewisham Merton Newham Redbridge Richmond upon Thames Southwark Sutton Tower Hamlets Waltham Forest Wandsworth Westminster

NCARDRS region	Local authorities	
Northern	Allerdale Carlisle Copeland County Durham Darlington Eden Gateshead Hartlepool	Middlesbrough Newcastle upon Tyne North Tyneside Northumberland Redcar and Cleveland South Tyneside Stockton-On-Tees Sunderland
North West	Halton Warrington Blackburn with Darwen Blackpool Cheshire East Cheshire West and Chester Barrow-in-Furness South Lakeland Burnley Chorley Fylde Hyndburn Lancaster Pendle Preston Ribble Valley Rossendale South Ribble	West Lancashire Wyre Bolton Bury Manchester Oldham Rochdale Salford Stockport Tameside Trafford Wigan Knowsley Liverpool St.Helens Sefton Wirral
South West	Bath and North East Somerset Bristol, City of Cheltenham Cornwall Cotswold East Devon Exeter Forest of Dean Gloucester Isles of Scilly Mendip Mid Devon North Devon	Plymouth Sedgemoor South Gloucestershire South Hams South Somerset Stroud Swindon Taunton Deane Teignbridge Tewkesbury Torbay Torridge West Devon West Somerset

NCARDRS region	Local authorities	
	North Somerset	Wiltshire (excluding Salisbury)
Thames Valley	Aylesbury Vale Bracknell Forest Cherwell Chiltern Milton Keynes Oxford Reading Slough	South Bucks South Oxfordshire Vale of White Horse West Berkshire Windsor and Maidenhead Wokingham Wycombe
Wessex	Basingstoke and Deane Bournemouth Christchurch East Dorset East Hampshire Eastleigh Fareham Gosport Hart Havant Isle of Wight New Forest	North Dorset Poole Portsmouth Purbeck Rushmoor Southampton Test Valley West Dorset Weymouth and Portland Wiltshire (Salisbury only) Winchester
West Midlands	Birmingham Bromsgrove Cannock Chase Coventry Dudley East Staffordshire Herefordshire, County of Lichfield Malvern Hills Newcastle-under-Lyme North Warwickshire Nuneaton and Bedworth Redditch Rugby Sandwell	Shropshire Solihull South Staffordshire Stafford Stoke-on-Trent Stratford-on-Avon Tamworth Telford and Wrekin Walsall Warwick Wolverhampton Worcester Wychavon Wyre Forest

NCARDRS region	Local authorities	
Yorkshire and Humber	Bradford Calderdale Craven East Riding of Yorkshire Hambleton Harrogate Kingston upon Hull, City of Kirkless	Leeds Richmondshire Ryedale Scarborough Selby Wakefield York

Data collection

Congenital anomalies are defined as being present at delivery, originating before birth, and include structural, chromosomal and genetic anomalies. Screening during pregnancy can detect some congenital anomalies, while some are found at birth. Others are detected 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 nationally and ensuring consistency and standardisation across the country.

A single data management system has been developed and NCARDRS has a growing team of 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 TOPFA at any gestation and miscarriages where an anomaly is present is also collected. NCARDRS only report anomalies that are recorded in pregnancies that end in a late miscarriage (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 registrations and for the validation of cases, ensuring accurate matching between antenatally diagnosed anomalies and postnatal notifications.

Data quality

All 10 reporting regions have submitted data to the European Surveillance of Congenital Anomalies (EUROCAT) since the 2018 birth year cohort and followed their data quality procedures, ensuring collection of a number of core variables. More information can be found in the EUROCAT Guidelines for data registration. 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) greater than or equal to 20 weeks and TOPFA (at any gestational age) with at last one registered anomaly delivered in England are included for reporting.

Coding and reporting

NCARDRS codes congenital anomalies according to the paediatric adaptation of ICD-10 produced by the British Paediatric Association (BPA). The BPA classification specifies more clinical terms than ICD-10, and provides greater granularity for analytical purposes through the use of 5th character extensions to many ICD-10 codes. NCARDRS uses the EUROCAT congenital anomaly subgroup categories for reporting. These subgroups use ICD10 codes with the BPA extension to group together conditions by body system and anomaly type. Table 2 reproduces the EUROCAT congenital anomaly subgroup categories. Table 3 lists the exclusions applied to the categories in Table 2.

Table 2. EUROCAT congenital anomaly subgroups

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
All anomalies	Q-chapter, D21.5, D82.1, D18.10, P35.0, P35.1, P37.1		Exclude all minor anomalies as specified in exclusion list below
Nervous system	Q00*, Q01*, Q02, Q03*, Q04*, Q05*, Q06*, Q07*		Q04.61, Q07.82
Neural tube defects	Q00*, Q01*, Q05*		
Anencephalus and similar	Q00*		
Encephalocele	Q01*	Exclude if associated with anencephalus subgroup	
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 or holoprosencephaly	Q04.1, Q04.2		
Eye	Q10*-Q15*		Q10.1-Q10.3, Q10.5, Q13.5
Anophthalmos or microphthalmos	Q11.0, Q11.1, Q11.2		
Anophthalmos	Q11.0, Q11.1		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Congenital cataract	Q12.0		
Congenital glaucoma	Q15.0		
Ear, face and neck	Q16*, Q17*, Q18*		Q17.0-Q17.5, Q17.9, Q18.0-Q18.2, Q18.4-Q18.7, Q18.80, Q18.9
Anotia	Q16.0		
Congenital Heart Defects	Q20*-Q26*	Exclude Patent ductus arteriosuswith gestational age (GA) less than 37 weeks Exclude peripheral pulmonary artery stenosis with GA less than 37 weeks	Q21.11, Q25.0 if GA less than 37 weeks, Q25.41, Q25.6 if GA less than 37 weeks, Q26.1
Severe CHD	Q20.0, Q20.1, Q20.3, Q20.4, Q21.2, Q21.3, Q22.0, Q22.4, Q22.5, Q22.6, Q23.0, Q23.2, Q23.3, Q23.4, Q25.1, Q25.2, Q26.2		
Common arterial truncus	Q20.0		
Double outlet right ventricle	Q20.1		
Transposition of great vessels	Q20.3		
Single ventricle	Q20.4		
Ventricular septal defect	Q21.0		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Atrial septal defect	Q21.1		Q21.11
Atrioventricular septal defect	Q21.2		
Tetralogy of Fallot	Q21.3		
Tricuspid atresia and stenosis	Q22.4		
Ebstein's anomaly	Q22.5		
Pulmonary valve stenosis	Q22.1		
Pulmonary valve atresia	Q22.0		
Aortic valve atresia or stenosis	Q23.0		
Mitral valve anomalies	Q23.2, Q23.3		
Hypoplastic left heart	Q23.4		
Hypoplastic right heart	Q22.6		
Coarctation of aorta	Q25.1		
Aortic atresia or interrupted aortic arch	Q25.2		
Total anomalous pulm venous return	Q26.2		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Patent ductus arteriosus as **only** CHD in term infants (GA +37 weeks)	Q25.0	Livebirths only	
Respiratory	Q30.0, Q32*-Q34*	Exclude Q33.6	Q32.0, Q33.1
Choanal atresia	Q30.0		
Cystic adenomatous malformation of lung	Q33.80		
Oro-facial clefts	Q35*-Q37*	Exclude association with holoprosencephaly or anencephaly subgroups	
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*, Q79.0		Q38.1, Q38.2, Q38.50, Q40.0, Q40.1. Q40.21, Q43.0, Q43.20, Q43.81, Q43.82
Oesophageal atresia with or without tracheo-oesophageal fistula	Q39.0-Q39.1		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Duodenal atresia or stenosis	Q41.0	Exclude if also annular pancreas subgroup	
Atresia or stenosis of other parts of small intestine	Q41.1-Q41.8		
Ano-rectal atresia and stenosis	Q42.0-Q42.3		
Hirschsprung's disease	Q43.1		
Atresia of bile ducts	Q44.2		
Annular pancreas	Q45.1		
Diaphragmatic hernia	Q79.0		
Abdominal wall defects	Q79.2, Q79.3, Q79.5		
Gastroschisis	Q79.3		
Omphalocele	Q79.2		
Urinary	Q60*-Q64*, Q79.4		Q61.0, Q62.7, Q63.3
Bilateral renal agenesis including Potter syndrome	Q60.1, Q60.6	Exclude unilateral	
Multicystic Renal dysplasia	Q61.40, Q61.41		
Congenital hydronephrosis	Q62.0		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Bladder exstrophy and/or epispadias	Q64.0, Q64.1		
Posterior urethral valve and/or prune belly	Q64.20, Q79.4		
Genital	Q50*-Q52*, Q54*-Q56*		Q52.3, Q52.5, Q52.7, Q55.20, Q55.21
Hypospadias	Q54*		
Indeterminate sex	Q56*		
Limb	Q65*-Q74*		Q65.3-Q65.6, Q66.2-Q66.9, Q67.0-Q67.8, Q68.0, Q68.10, Q68.21, Q68.3-Q68.5, Q74.00
Limb reduction	Q71*-Q73*		
Club foot – talipes equinovarus	Q66.0		
Hip dislocation and/or dysplasia	Q65.0–Q65.2, Q65.80, Q65.81		
Polydactyly	Q69*		
Syndactyly	Q70*		
Other anomalies or syndromes			
Skeletal dysplasias	Q74.02, Q77*, Q78.00, Q78.2-Q78.8		
Craniosynostosis	Q75.0		

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Congenital constriction bands or amniotic band	Q79.80		
Situs inversus	Q89.3		
Conjoined twins	Q89.4		
Congenital skin disorders	Q80*-Q82*		Q82.5, Q82.80
VATER/VACTERL	Q87.26		
Vascular disruption anomalies	Q04.35, Q41.1, Q41.2, Q41.8, Q71.0, Q71.2, Q71.3, Q72.0, Q72.2, Q72.3, Q73.0, Q79.3, Q79.5, Q79.80, Q79.82, Q87.06		
Laterality anomalies	Q20.6, Q24.0, Q33.81, Q89.0, Q89.3		
Teratogenic syndromes with malformations	Q86*, P35.0, P35.1, P37.1		
Fetal alcohol syndrome	Q86.0		
Valproate syndrome	Q86.80		
Maternal infections resulting in malformations	P35.0, P35.1, P37.1		
Genetic syndromes + microdeletions	Q44.71, Q61.90, Q74.84, Q75.1, Q75.4, Q75.81, Q87*, Q93.6, D82.1	Exclude Associations and sequences Exclude Q87.03, Q87.04, Q87.06, Q87.08, Q87.24, Q87.26	

Subgroups	ICD10-BPA	Comments	Excluded minor anomalies
Chromosomal	Q90*-Q92*, Q93*, Q96*-Q99*	Exclude microdeletions Q93.6	
Down's syndrome	Q90*		
Patau's syndrome	Q91*4-Q91*7		
Edwards' syndrome	Q91*0-Q91*3		
Turner's syndrome	Q96*		
Klinefelter's syndrome	Q98*0-Q98*4		

Table 3. List of exclusions

	Specified ICD10-BPA – if present
Head	
Aberrant scalp hair patterning	
Brachycephaly	
Flat occiput	
Depressions in skull, lacunar skull, temporal flattening	Q67.40
Dolichocephaly	Q67.2
Plagiocephaly – head asymmetry	Q67.3
Third fontanelle	
Macrocephalus	Q75.3
Facial asymmetry	Q67.0
Compression facies	Q67.1
Other cong deformities of skull, face and jaw (including all types of abnormally shaped skull without synostosis)	Q67.4
Skull, late closure	
Dysmorphic face Broad, prominent forehead	Q18.9
Coarse facies	
Flattened face	
Frontal bossing or 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	H04.6
Epicanthic folds	Q18.9
Epicenthus inversus	Q18.9

	Specified ICD10-BPA – if present
Exophthalmos	H05.2
Upward slanting palpebral fissures	Q10.3
Downward slanting palpebral fissures	Q10.3
Short palpebral fissures	Q18.9
Congenital ectropion	Q10.1
Congenital entropion	Q10.2
Other congenital malformations of eyelid	Q10.3
Oval shaped pupils	
Prominent or protruding eyes	H05.2
Dystopia canthorum	Q18.9
Hypertelorism	Q75.2
Hypotelorism	Q18.9
Stenosis of stricture of lacrimal duct	Q10.5
Synophrys	Q18.80
Blue sclera	Q13.5
Crocodile tears	Q07.82
Ears	
Primitive shape	Q17.3
Lack of helical fold	Q17.3
Asymmetric size	Q17.3
Posterior angulation	Q17.3
Microtia or small ears	Q17.2
Macrotia	Q17.1
Protuberant ears	Q17.3
Absent tragus	
Double lobule	Q17.0
Accesorry auricle, preauricular appendage, tag or lobule	Q17.0
Auricular pit	
Pointed ear, Vulcan ear, simple ear	Q17.3
Preauricular sinus or cyst	Q18.1
Narrow external auditory meatus	
Low set ears	Q17.4

	Specified ICD10-BPA – if present
Bat ear, prominent proturberant ear	Q17.5
Congenital absence of ear lobe	
Darwin's tubercle	
Unspecified and minor malformation of ear	Q17.9
Nose	
Small or hypoplastic nares	Q18.9
Notched alas	
Anteverted nares	Q18.9
Bifid tip of nose	Q18.9
Broad nasal root, anomaly of nasal root	Q18.9
Depressed nasal bridge	Q18.9
Deviation of nasal septum	Q67.41
Dysmorphic nose	Q18.9
Flat nose	Q18.9
Flattened nasal bridge	Q18.9
Pinched nose	Q18.9
Prominent nasal bridge	Q18.9
Saddle nose	Q18.9
Small pointed nose	Q18.9
Underdeveloped nasal bones	Q18.9
Upturned nose	Q18.9
Wide nasal root	Q18.9
Oral regions	
Borderline small mandible or minor micrognathia	
Aberrant frenula	
Absentor hypoplasia depressor anguli oris (asymmetric crying face)	
Alveolar crest	
Anomalies of philtrum, elongated philtrum	Q18.9
Bifid uvula or cleft uvula	Q35.7
Borderline small mandible or minor micrognathia	
Disturbances in tooth eruption	

	Specified ICD10-BPA – if present
Enamel hypoplasia	
Glossoptosis	
Malformed teeth	
High arched palate	Q38.50
Tongue tie or cyst of tongue	Q38.1
Macroglossia or hemi-hypertrophy of tongue	Q38.2
Macrostomia	Q18.4
Malformed teeth	
Microstomia	Q18.5
Macrocheilia	Q18.6
Microcheilia	Q18.7
Microglossia	
Microstomia	Q18.9
Mid-oral tongue position	
Neonatal teeth	
Prominent jaw	Q18.9
Retrognathia or receding chin	Q67.4
Short philtrum	Q18.9
Thin lips	Q18.9
Ranula	
Neck	
Broad neck	Q18.9
Congenital thymic hypoplasia	
Short neck	Q18.9
Mild webbed neck	
Sinus, fistula or cyst of branchial cleft	Q18.0
Thymus involution	
Thyreoglossal cyst	
Preauricular sinus or cyst	Q18.1
Other branchial cleft malformations	Q18.2
Congenital malformation of face and neck, unspecified	Q18.9
Torticollis	Q68.0

	Specified ICD10-BPA – if present
Hands	
Duplication of thumbnail	
Arachnodactyly	
Enlarged or hypertrophic nails	Q84.5
Single or abnormal palmar crease	Q82.80
Unusual dermatoglyphics	
Clinodactyly (5th finger)	Q68.10
Short fingers (4. 5. th finger)	
Accessorry carpal bones	Q74.00
Other congenital malformations of nails	Q84.6
Overlapping fingers	
Small fingers	
Subluxation of phalangeal bones	
Feet or limb	
Bulbous toes	
Syndactyly (second to third toes)	
Gap between toes (first to second)	
Short great toe	
Recessed toes (fourth, fifth)	
Enlarged or hypertrophic nails	Q84.5
Prominent calcaneus	
Clicking hip, subluxation of unstable hip	Q65.3-Q65.6
Metatarsus varus or metatarsus adductus	Q66.2
Hallux varus – other cong varus deformities of feet	Q66.3
Talipes or pes calcaneovalgus	Q66.4
Talipes calcaneovarus	Q66.1
Congenital pes planus	Q66.5
Hip dysplasia and other specified or unspecified hip anomalies	Q65.8, Q65.9
Metatarsus varus – other cong valgus deformities of feet	Q66.6
Overlapping toes	
Pes cavus	Q667

	Specified ICD10-BPA – if present
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	Q82.5
Neavus flammeus	Q82.50
Strawberry naevus	Q82.51
Lymphangioma if no treatment is required	
Angioma	
Persistent lanugo	
Mongoloid spot (whites)	Q82.52
Depigmented spot	
Unusual placement of nipples or wide spaced nipples	
Accessory nipples	Q83.3
Accessory skin tags	Q82.81
Cafe-au-lait spot	
Epibulbar dermoid	
Heterochromia of hair	
Hypoplasia of toe nails	Q84.6
Skeletal	
Cubitus valgus	
Prominent sternum or pectus carinatum	Q67.7
Prominent sternum	
Depressed sternum or pectus excavatum	Q67.6
Sternum bifidum	Q76.71
Shieldlike chest, other cong deformities of chest	Q67.8
Congenital deformity of spine	Q67.5
Genua valgum	
Genus varum	
Genu recurvatum	Q68.21

	Specified ICD10-BPA – if present
Duplication of ribs	
Congenital bowing of femur	Q68.3
Congenital bowing of fibula and tibia	Q68.4
Congenital bowing of long bones of leg, unspecified	Q68.5
Congenital bowing of upper limb	
No ossification of os coccyx	
Ovoid configuration of vertebrae	
Spina bifida occulta	Q76.0
Sacral dimple	L05.9
Cervical rib	Q76.5
Fused rib, single	
Absence of rib or hypoplastic rib	Q76.60
Accessory rib	Q76.62
Congenital lordosis, postural	Q76.43
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	Q07.80
Periventricular leukomalacia	
Single congenital cerebral cyst	Q04.61
Thin or hypoplastic corpus callosum	
Ventriculomegaly less than 15 mm	
Choroid plexus cysts	

	Specified ICD10-BPA – if present
Anomalies of septum pellucidum	
Cardiovascular	
Absence or hypoplasia of umbilical artery, single umbilical artery	Q27.0
Absence of vena cava superior	
Functional or unspecified cardiac murmur	R01.1
Cardiomegaly	I51.7
Cardiomyopathy	I42.9
Deviation of the heart axis	
Patent ductus arteriosus if GA less than 37 weeks	Q25.0 if GA less than 37 weeks
Peripheral pulmonary artery stenosis	Q25.6 if GA less than 37 weeks
Patent or persistent foramen ovale	Q21.11
Persistent left superior vena cava	Q26.1
Persistent right aortic arch	Q25.41
Persistent right umbilical vein	
Congenital heart block	Q24.6
Pulmonary	
Accessory lobe of lung	Q33.1
Congenital laryngeal stridor	Q31.4
Laryngomalacia	Q31.40
Tracheomalacia	Q32.0
Azygos lobe of lung	Q33.10
Bronchomalacia	Q32.2
Single cyst of the lung	Q33.00
Hyperplasia of thymus	
Pleural effusion	
Pulmonary hypoplasia, secondary	
Relaxation of diaphragm	
Thymus involution	
Vocal cord palsy	
Gastro-intestinal	
Hiatus hernia	Q40.1

	Specified ICD10-BPA – if present
Abdominal cyst not needing surgery	
Accessory spleen	
Choledochal cyst	Q44.4
Congenital adrenal hypoplasia	Q89.11
Congenital cholestasis	
Congenital mesenteric cyst	Q45.83
Cyst of spleen	
Dilatation of intestine	
Hepatomegaly	R16.0
Liver cyst	
Plica of anus	
Splenomegaly	
Pyloric stenosis	Q400
Diastasis recti	
Umbilical hernia	
Inguinal hernia	K40.9
Meckel's diverticulum	Q43.0
Functional gastro-intestinal disorders	Q40.21, Q43.20, Q43.81, Q43.82
Transient choledochal cyst	
Anterior anus without surgery	
Renal	
Vesico-ureteral-renal reflux	Q62.7
Enlarged or thickened bladder	
Hydronephrosis with a pelvis dilatation less than 10 mm	
Hyperplastic and giant kidney	Q63.3
Single renal cyst	Q61.0
External genitals	
Deficient or hooded foreskin	
Undescended testicle	Q53*
Unspecified ectopic testis	
Retractile testis	Q55.20
Hydrocele of testis	

	Specified ICD10-BPA – if present
Phymosis	
Bifid scrotum	Q55.21
Buried penis	
Congenital chordee	Q54.4
Congenital adrenogenital disorders	E25.0
Congenital malformation of vulva	Q52.7
Congenital torsion of ovary	Q50.2
Curvature of penis	
Hypoplasia of penis or micropenis	
Hymen imperforate	Q52.3
Fusion of labia	Q52.5
Prominent labia minora	
Enlarged clitoris	
Vaginal skin tag	
Cysts of vulva	
Transient ovarian cyst	
Developmental ovarian cyst(s)	Q50.1, Q50.10, Q50.11
Embryonic cyst of broad ligament	Q50.5
Foreskin tethered to the scrotum	N47
Hypertrophy of hymen	
Phimosis	N47
Seminal vesicle cyst	
Testicular torsion	N44
Other	
Congenital malformation, unspecified	Q89.9
Chromosomal	
Balanced chromosomal rearrangements	Q95*
Balanced translocations or inversions in normal individuals	
Balanced autosomal rearrangement in abnormal individual	Q95.2
Individuals with marker heterochromatin	
Individuals with autosomal fragile site	

Down's syndrome, Edwards' syndrome and Patau's syndrome

Source data and completeness

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.

Inclusion and exclusion criteria

All babies with Down's syndrome, Edwards' syndrome or Patau's syndrome delivered in 2019 according to the case definition previously described 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). Babies with a positive non-invasive prenatal testing (NIPT) and a clinical suspicion of Down's syndrome, Edwards' syndrome or Patau's syndrome based on postnatal phenotype, but with no further testing, are also included in this report.

Reference

Bégaud B, Martin K, Abouelfath A, Tubert-Bitter P, Moore N, Moride Y. ‘An easy to use method to approximate Poisson confidence limits.’ European Journal of Epidemiology 2005: volume 20, pages 213-216

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.

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