



Public Health
England

Protecting and improving the nation's health

National Congenital Anomaly and Rare Disease Registration Service

Congenital anomaly statistics 2016 – glossary and technical details

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Glossary of terms

Term	Definition
Antenatal	The period from conception to birth.
Antenatal diagnosis	A diagnosis made in a live fetus at any gestation.
Birth prevalence	The total number of cases of congenital anomaly (live births, stillbirths, late miscarriages and terminations of pregnancy with fetal anomaly) compared to the total number of births (live births and stillbirths). ⁱ
Births/total births	Live births and stillbirths. ⁱⁱ
Case	A baby/fetus with one or more congenital anomaly. Includes live births, stillbirths, late miscarriages and terminations of pregnancy with fetal anomaly.
Case ascertainment	Proportion of notifications of congenital anomalies reported to NCARDRS out of all cases of congenital anomaly in the population.
Confidence interval (see below for more information)	The range (confidence interval) about the sample proportion/mean will actually contain the true - but unknown - proportion/mean value in the population. It is usual practice to create confidence intervals at the 95% level which means 95% of the time our confidence intervals should contain the true value found in the population.
Congenital anomaly	Present at delivery, probably originating before birth, and includes structural, chromosomal, genetic and biochemical malformations.
Congenital hydronephrosis	An obstruction of the urinary flow from kidney to bladder. Cases are registered where the renal pelvis measurement is ≥ 10 mm after birth. ⁱ
Detection rate	The proportion of affected individuals with a positive screening result.
Feticide	A procedure to stop the fetal heart and cause the demise of the fetus in the uterus. ⁱⁱⁱ
Genetic anomalies	Includes chromosomal, skeletal dysplasias, genetic syndromes and microdeletions ⁱ

Infant deaths	Deaths under 1 year of age. ⁱⁱ
Infant mortality	The number of infant deaths per 10,000 live births.
Late miscarriage	Late fetal deaths from 20-23 completed weeks of gestation.
Live birth	A baby showing signs of life at birth. ⁱⁱ
Major congenital anomaly subgroup (see below for more information)	The high level body system and anomaly type groupings of congenital anomalies. ⁱ
Perinatal deaths	Stillbirths and deaths under 7 days of age. ⁱⁱ
Perinatal mortality	The number of perinatal deaths per 10,000 total births.
Severe congenital heart defects (CHD)	This includes the following congenital heart defects ⁱ : <ul style="list-style-type: none"> • Common arterial truncus • Transposition of great vessels • Single ventricle • Atrioventricular septal defect • Tetralogy of Fallot • Tricuspid atresia and stenosis • Ebstein's anomaly • Pulmonary valve atresia • Aortic valve atresia/stenosis • Hypoplastic left heart • Hypoplastic right heart • Coarctation of aorta • Total anomalous pulmonary venous return
Severe microcephaly	Where the head circumference is less than - 3 standard deviations for sex and gestational age. ⁱ
Stillbirths	A baby born after 24 or more weeks completed gestation and which did not, at any time, breathe or show signs of life. ⁱⁱ
Statistical significance (see below for more information).	Statistical testing is undertaken by comparing the confidence intervals to see if they overlap - with non-overlapping confidence intervals being considered as statistically significantly different.
Termination of pregnancy with fetal anomaly (TOPFA)	Term used to describe the deliberate ending of a pregnancy with the intention that the fetus will not survive and which is carried out when the fetus is diagnosed prenatally as having a major congenital anomaly.

	<p>This includes terminations of pregnancy for fetal anomaly as well as terminations of pregnancy for other medical reasons where a fetal anomaly was present.</p> <p>Where a pregnancy ends in a TOPFA, the baby may be born dead, or if parents have not opted for prior feticide the baby may be born alive but die shortly after. Depending on the gestation at which a TOPFA takes place (before or after 24 weeks) it may also be registered as a stillbirth.</p>
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ⁱ <http://www.euocat-network.eu/>

ⁱⁱ <https://www.ons.gov.uk/>

ⁱⁱⁱ Royal College of Obstetricians and Gynaecologists. The Future Workforce in Obstetrics and Gynaecology in England and Wales. Working Party Report. London: RCOG; 2009.

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’ cases of 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 with 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 ‘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 two 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.ⁱⁱⁱ 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.ⁱⁱⁱ

Calculation of birth prevalence and their 95% confidence intervals

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

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

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

Data collection

A congenital anomaly is defined as any defect present at delivery, probably originating before birth, and includes structural, chromosomal, genetic and biochemical defects and malformations. 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 is 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 live births and stillbirths affected by congenital anomalies, information about terminations of pregnancy with a diagnosed fetal

anomaly at any gestation (TOPFA) and miscarriages where an anomaly is present, are also collected. NCARDRS only report on late miscarriages (20-23 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 validations of cases, ensuring accurate matching between antenatally diagnosed anomalies and postnatal notifications.

Data quality

All seven reporting regions submitted data to the [European Surveillance of Congenital Anomalies](#) (EUROCAT) and followed their data quality procedures. 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.

Reporting

NCARDRS use 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		Q0782
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	
Microcephaly	Q02	Exclude association with NTD subgroup	
Arhinencephaly / holoprosencephaly	Q041, Q042		
Congenital Heart Defects	Q20-Q26	Exclude isolated PDA with GA<37 weeks	Q2111, Q250 if GA <37 weeks
Severe CHD	Q200, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q234, Q251, 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	Q30-Q34		Q314, Q315, Q320, Q331
Choanal atresia	Q300		
Cystic adenomatous malformation of lung	Q3380		
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, 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	
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
Renal dysplasia	Q614	
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, Q5520, Q5521
Hypospadias	Q54	
Indeterminate sex	Q56	
Limb	Q65-Q74	Q653-Q656, Q662-Q669, Q670-Q678, Q680, 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, Q8716	
Genetic syndromes + microdeletions	Q4471, Q6190, Q7484, Q751, Q754, Q7581, Q87, Q936, D821 Exclude Q8703, Q8704, Q8706, Q8708, Q8724, Q8726	Exclude Associations and sequences
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	
Flat occiput	
Dolichocephaly	Q67.2
Plagiocephaly – head asymmetry	Q67.3
Bony occipital spur	
Third fontanel	
Macrocephalus	Q75.3
Facial asymmetry	Q67.0
Compression facies	Q67.1
Other cong deformities of skull, face and jaw	Q67.4
Eyes	
Epicanthic folds	
Epicenthus inversus	
Upward slanting palpebral fissures	
Downward slanting palpebral fissures	
Short palpebral fissures	
Congenital ectropion	Q10.1
Congenital entropion	Q10.2
Other congenital malformation of eyelid	Q10.3
Dystopia canthorum	
Hypertelorism	Q75.2
Hypotelorism	
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	Q17.2
Macrotia	Q17.1
Protuberant ears	Q17.3
Absent tragus	
Double lobule	Q17.0
Accessory auricle, preauricular appendage, tag or lobule	Q17.0
Auricular pit	
Preauricular sinus or cyst	Q18.1
Narrow external auditory meatus	
Low set ears	Q17.4
Bat ear, prominent ear	Q17.4
Unspecified and minor malformation of ear	Q17.9
Nose	
Small nares	

Notched alas	
Oral regions	
Borderline small mandible/ minor micrognathia	
Aberrant frenula	
Enamel hypoplasia	
Malformed teeth	
High arched palate	Q38.50
Tongue tie or cyst of tongue	Q38.1
Macroglossia	Q38.2
Macrostomia	Q18.4
Microstomia	Q18.5
Macrocheilia	Q18.6
Microcheilia	Q18.7
Ranula	
Neck	
Mild webbed neck	
Sinus, fistula or cyst of branchial cleft	Q18.0
Preauricular sinus or cyst	Q18.1
Other branchial cleft malformation	Q18.2
Congenital malformation of face and neck, unspecified	Q18.9
Torticollis	Q68.0
Hands	
Duplication of thumbnail	
Enlarged or hypertrophic nails	Q84.5
Single/abnormal palmar crease	Q82.80
Unusual dermatoglyphics	
Clinodactyly (5th finger)	
Short fingers (4. 5. th finger)	
Accessory carpal bones	Q74.00
Feet -Limb	
Syndactyly (2nd-3rd toes)	
Gap between toes (1st-2nd)	
Short great toe	
Recessed toes (4th, 5th)	
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
Congenital pes planus	Q66.5
Metatarsus varus – other cong valgus deformities of feet	Q66.6
Pes cavus	Q66.7
Clubfoot of postural origin – other cong deformities of feet	Q66.8
Congenital deformity of feet, unspecified	Q66.9
Skin	
Hemangioma (other than face or neck)	
Pigmented naevus – cong non-neoplastic naevus	Q82.5
Neavus flammeus	Q82.50
Strawberry naevus	Q82.51
Lymphangioma	
Angioma	
Persistent lanugo	
Mongoloid spot (whites)	Q82.52
Depigmented spot	
Unusual placement of nipples	
Accessory nipples	Q83.3
Cafe-au-lait spot	

Skeletal	
Cubitus valgus	
Prominent sternum	Q67.7
Depressed sternum	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
Congenital bowing of femur	Q68.3
Congenital bowing of fibula and tibia	Q68.4
Congenital bowing of long bones of leg, unspecified	Q68.5
Spina bifida occulta	Q76.0
Sacral dimple	
Cervical rib	Q76.5
Absence of rib	Q76.60
Accessory rib	Q76.62
Congenital lordosis, postural	Q76.43
Brain	
Arachnoid cyst	
Choroid plexus cyst	
Anomalies of septum pellucidum	
Cardiovascular	
Absence or hypoplasia of umbilical artery, single umbilical artery	Q27.0
Functional or unspecified cardiac murmur	
Patent ductus arteriosus if GA < 37 weeks	Q25.0 if GA <37 weeks
Peripheral pulmonary artery stenosis	
Patent or persistent foramen ovale	Q21.11
Pulmonary	
Accessory lobe of lung	Q33.1
Congenital laryngeal stridor	Q31.4
Laryngomalacia	Q31.4, Q31.5
Tracheomalacia	Q32.0
Azygos lobe of lung	Q33.10
Gastro-intestinal	
Hiatus hernia	Q40.1
Pyloric stenosis	Q40.0
Diastasis recti	
Umbilical hernia	
Inguinal hernia	
Meckel's diverticulum	Q43.0
Functional gastro-intestinal disorders	Q40.21, Q43.20, Q43.81, Q43.82
Transient choledochal cyst	
Anterior anus	
Renal	
Vesico-ureteral-renal reflux	Q62.7
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	
Phymosis	
Bifid scrotum	Q55.21

Curvature of penis lateral	
Hypoplasia of penis	
Hymen imperforatum	Q52.3
Fusion of labia	Q52.5
Prominent labia minora	
Enlarged clitoris	
Vaginal skin tag	
Cysts of vulva	
Transient ovarian cyst	
Other	
Congenital malformation, unspecified	Q89.9
Chromosomal	
Balanced translocations or inversions in normal individuals	Q95.0, Q95.1

'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

Fetal Anomaly Screening Programme (FASP) anomalies

Anomaly	ICD-10 BPA	FASP Category
Anencephaly	Q00	Anencephaly
Spina bifida	Q05	Spina bifida
Cleft lip	Q36	Cleft lip
Cleft lip and palate	Q37	Cleft lip
Transposition of great vessels	Q203	Serious cardiac
Atrioventricular septal defect	Q212	Serious cardiac
Tetralogy of Fallot	Q213	Serious cardiac
Pentalogy of Fallot	Q2182	Serious cardiac
Hypoplastic left heart	Q234	Serious cardiac
Bilateral renal agenesis	Q601	Bilateral renal agenesis
Achondrogenesis	Q770	Lethal skeletal dysplasia
Thanatophoric short stature	Q771	Lethal skeletal dysplasia
Short Rib Syndrome	Q772	Lethal skeletal dysplasia
Other osteochondrodysplasia with defect of growth of tubular bones and spine	Q779	Lethal skeletal dysplasia
Osteogenesis imperfecta	Q780	Lethal skeletal dysplasia
Congenital diaphragmatic hernia	Q790	Congenital diaphragmatic hernia

Exomphalos	Q792	Exomphalos
Gastroschisis	Q793	Gastroschisis
Down's syndrome	Q90	Down's syndrome
Edward's syndrome	Q910-Q913	Edward's syndrome
Patau's syndrome	Q914-917	Patau's syndrome

Comparison of EUROCAT and FASP inclusion criteria

EUROCAT	FASP
Deliveries in 2016, regardless of gestation at delivery	EDD between 01/04/2015 and 31/03/2016, regardless of date of delivery
Based on mother's residence at delivery	Based on initial booking hospital
All outcomes included except fetal losses under 20 weeks' gestation	All outcomes of pregnancy included
Minimum data required includes date of delivery, postcode and at least one anomaly from EUROCAT inclusion list	Minimum data required includes booking hospital, EDD, at least one anomaly from FASP inclusion list and sufficient detail on screen pathway to categorise detection

Geographical coverage of the NCARDRS regions in this report

NCARDRS region

East Midlands and South Yorkshire

Local Authorities

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

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
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 North Somerset	Plymouth Sedgemoor South Gloucestershire South Hams South Somerset Stroud Swindon Taunton Deane Teignbridge Tewkesbury Torbay Torrige West Devon West 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 & 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

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