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# National Congenital Anomaly and Rare Disease Registration Service

## Congenital anomaly statistics 2016 – tables

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Published October 2018
PHE publications
gateway number: 2018527

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# Table 1: Number of cases, birth prevalence (per 10,000 total births) and 95% confidence intervals according to congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | Including genetic[2] | | | | | | Excluding genetic[2] | |
|---|---|---|---|---|---|---|---|---|
| | Live births | Stillbirths[3] | Miscarriages[4] | TOPFA[5] | Total | Prevalence (95% CI) | Total | Prevalence (95% CI) |
| **Total births** | **327,900** | **1,401** | **z** | **z** | **329,301** | **z** | **z** | **z** |
| **All cases** | **4,867** | **191** | **77** | **1,617** | **6,752** | **205.0 (200.2-210.0)** | **4,968** | **150.9 (146.7-155.1)** |
| **Nervous system** | **299** | **24** | **8** | **446** | **777** | **23.6 (22.0-25.3)** | **691** | **21.0 (19.4-22.6)** |
| Neural tube defects | 102 | 10 | 6 | 307 | 425 | 12.9 (11.7-14.2) | 412 | 12.5 (11.3-13.8) |
| Anencephalus and similar | 18 | 2 | 4 | 160 | 184 | 5.6 (4.8-6.5) | 180 | 5.5 (4.7-6.3) |
| Encephalocele | 8 | 2 | 0 | 27 | 37 | 1.1 (0.8-1.5) | 33 | 1.0 (0.7-1.4) |
| Spina bifida | 76 | 6 | 2 | 120 | 204 | 6.2 (5.4-7.1) | 199 | 6.0 (5.2-6.9) |
| Hydrocephalus | 107 | 6 | 0 | 63 | 176 | 5.3 (4.6-6.2) | 153 | 4.6 (3.9-5.4) |
| Microcephaly | 20 | 2 | 0 | 5 | 27 | 0.8 (0.5-1.2) | 25 | 0.8 (0.5-1.1) |
| Arhinencephaly/holoprosencephaly | 10 | 1 | 0 | 40 | 51 | 1.5 (1.2-2.0) | 29 | 0.9 (0.6-1.3) |
| **Congenital heart defects (CHD)** | **1,812** | **59** | **19** | **279** | **2,169** | **65.9 (63.1-68.7)** | **1,732** | **52.6 (50.1-55.1)** |
| Severe CHD[6] | 635 | 29 | 7 | 194 | 865 | 26.3 (24.5-28.1) | 669 | 20.3 (18.8-21.9) |
| Common arterial truncus | 17 | 0 | 0 | 5 | 22 | 0.7 (0.4-1.0) | 15 | 0.5 (0.3-0.8) |
| Double outlet right ventricle | 56 | 3 | 0 | 26 | 85 | 2.6 (2.1-3.2) | 70 | 2.1 (1.7-2.7) |
| Transposition of great vessels | 89 | 1 | 1 | 21 | 112 | 3.4 (2.8-4.1) | 108 | 3.3 (2.7-4.0) |
| Single ventricle | 12 | 2 | 0 | 7 | 21 | 0.6 (0.4-1.0) | 20 | 0.6 (0.4-0.9) |
| Ventricular septal defect | 755 | 15 | 3 | 77 | 850 | 25.8 (24.1-27.6) | 678 | 20.6 (19.1-22.2) |
| Atrial septal defect | 488 | 12 | 3 | 24 | 527 | 16.0 (14.7-17.4) | 427 | 13.0 (11.8-14.3) |
| Atrioventricular septal defect | 132 | 8 | 2 | 45 | 187 | 5.7 (4.9-6.6) | 87 | 2.6 (2.1-3.3) |
| Tetralogy of Fallot | 116 | 4 | 1 | 22 | 143 | 4.3 (3.7-5.1) | 115 | 3.5 (2.9-4.2) |
| Tricuspid atresia and stenosis | 11 | 1 | 0 | 4 | 16 | 0.5 (0.3-0.8) | 16 | 0.5 (0.3-0.8) |
| Ebstein's anomaly | 10 | 3 | 0 | 2 | 15 | 0.5 (0.3-0.8) | 12 | 0.4 (0.2-0.6) |
| Pulmonary valve stenosis | 149 | 2 | 0 | 6 | 157 | 4.8 (4.1-5.6) | 145 | 4.4 (3.7-5.2) |
| Pulmonary valve atresia | 41 | 2 | 1 | 16 | 60 | 1.8 (1.4-2.3) | 50 | 1.5 (1.1-2.0) |
| Aortic valve atresia/stenosis | 52 | 3 | 0 | 6 | 61 | 1.9 (1.4-2.4) | 57 | 1.7 (1.3-2.2) |

| Congenital anomaly[1] | Including genetic[2] | | | | | | Excluding genetic[2] | |
|---|---|---|---|---|---|---|---|---|
| | Live births | Stillbirths[3] | Miscarriages[4] | TOPFA[5] | Total | Prevalence (95% CI) | Total | Prevalence (95% CI) |
| Mitral valve anomalies | 33 | 1 | 0 | 8 | 42 | 1.3 (0.9-1.7) | 39 | 1.2 (0.8-1.6) |
| Hypoplastic left heart | 45 | 2 | 0 | 47 | 94 | 2.9 (2.3-3.5) | 86 | 2.6 (2.1-3.2) |
| Hypoplastic right heart | 9 | 1 | 0 | 5 | 15 | 0.5 (0.3-0.8) | 13 | 0.4 (0.2-0.7) |
| Coarctation of aorta | 126 | 1 | 2 | 25 | 154 | 4.7 (4.0-5.5) | 128 | 3.9 (3.2-4.6) |
| Aortic atresia/interrupted aortic arch | 16 | 0 | 0 | 5 | 21 | 0.6 (0.4-1.0) | 16 | 0.5 (0.3-0.8) |
| Total anomalous pulmonary venous return | 29 | 0 | 0 | 3 | 32 | 1.0 (0.7-1.4) | 29 | 0.9 (0.6-1.3) |
| Patent ductus arteriosus as **only** CHD[7] in term infants (GA[8] 37+ weeks) | 79 | 0 | 0 | 0 | 79 | 2.4 (1.9-3.0) | 63 | 1.9 (1.5-2.4) |
| **Respiratory** | **111** | **8** | **6** | **33** | **158** | **4.8 (4.1-5.6)** | **139** | **4.2 (3.5-5.0)** |
| Choanal atresia | 20 | 0 | 0 | 4 | 24 | 0.7 (0.5-1.1) | 17 | 0.5 (0.3-0.8) |
| Cystic adenomatous malformation of lung | 59 | 0 | 0 | 3 | 62 | 1.9 (1.4-2.4) | 60 | 1.8 (1.4-2.3) |
| **Oro-facial clefts** | **456** | **5** | **2** | **48** | **511** | **15.5 (14.2-16.9)** | **454** | **13.8 (12.5-15.1)** |
| Cleft lip with or without cleft palate | 273 | 3 | 1 | 35 | 312 | 9.5 (8.5-10.6) | 284 | 8.6 (7.7-9.7) |
| Cleft palate | 183 | 2 | 1 | 13 | 199 | 6.0 (5.2-6.9) | 170 | 5.2 (4.4-6.0) |
| **Digestive system** | **474** | **20** | **6** | **100** | **600** | **18.2 (16.8-19.7)** | **497** | **15.1 (13.8-16.5)** |
| Oesophageal atresia with or without trachea-oesophageal fistula | 89 | 3 | 3 | 8 | 103 | 3.1 (2.6-3.8) | 95 | 2.9 (2.3-3.5) |
| Duodenal atresia or stenosis | 49 | 0 | 0 | 2 | 51 | 1.5 (1.2-2.0) | 22 | 0.7 (0.4-1.0) |
| Atresia or stenosis of other parts of small intestine | 29 | 1 | 0 | 1 | 31 | 0.9 (0.6-1.3) | 30 | 0.9 (0.6-1.3) |
| Ano-rectal atresia and stenosis | 93 | 1 | 1 | 33 | 128 | 3.9 (3.2-4.6) | 105 | 3.2 (2.6-3.9) |
| Hirschsprung's disease | 40 | 0 | 0 | 0 | 40 | 1.2 (0.9-1.7) | 35 | 1.1 (0.7-1.5) |
| Diaphragmatic hernia | 90 | 4 | 0 | 24 | 118 | 3.6 (3.0-4.3) | 105 | 3.2 (2.6-3.9) |
| **Abdominal wall defects** | **158** | **12** | **2** | **95** | **267** | **8.1 (7.2-9.1)** | **218** | **6.6 (5.8-7.6)** |
| Gastroschisis | 112 | 4 | 0 | 6 | 122 | 3.7 (3.1-4.4) | 121 | 3.7 (3.0-4.4) |
| Omphalocele | 42 | 7 | 2 | 74 | 125 | 3.8 (3.2-4.5) | 79 | 2.4 (1.9-3.0) |
| **Urinary** | **565** | **21** | **4** | **140** | **730** | **22.2 (20.6-23.8)** | **652** | **19.8 (18.3-21.4)** |
| Bilateral renal agenesis including Potter syndrome | 9 | 0 | 1 | 36 | 46 | 1.4 (1.0-1.9) | 42 | 1.3 (0.9-1.7) |
| Renal dysplasia | 121 | 2 | 0 | 27 | 150 | 4.6 (3.9-5.3) | 138 | 4.2 (3.5-5.0) |
| Congenital hydronephrosis | 168 | 5 | 0 | 10 | 183 | 5.6 (4.8-6.4) | 169 | 5.1 (4.4-6.0) |

| Congenital anomaly[1] | Including genetic[2] | | | | | | Excluding genetic[2] | |
|---|---|---|---|---|---|---|---|---|
| | Live births | Stillbirths[3] | Miscarriages[4] | TOPFA[5] | Total | Prevalence (95% CI) | Total | Prevalence (95% CI) |
| Bladder exstrophy and/or epispadias | 15 | 0 | 0 | 6 | 21 | 0.6 (0.4-1.0) | 20 | 0.6 (0.4-0.9) |
| Posterior urethral valve and/or prune belly | 44 | 0 | 0 | 5 | 49 | 1.5 (1.1-2.0) | 46 | 1.4 (1.0-1.9) |
| **Genital** | **374** | **5** | **2** | **22** | **403** | **12.2 (11.1-13.5)** | **369** | **11.2 (10.1-12.4)** |
| Hypospadias | 317 | 1 | 1 | 0 | 319 | 9.7 (8.7-10.8) | 308 | 9.4 (8.3-10.5) |
| Indeterminate sex | 18 | 0 | 0 | 4 | 22 | 0.7 (0.4-1.0) | 18 | 0.5 (0.3-0.9) |
| **Limb** | **633** | **34** | **13** | **124** | **804** | **24.4 (22.8-26.2)** | **697** | **21.2 (19.6-22.8)** |
| Limb reduction | 71 | 8 | 6 | 41 | 126 | 3.8 (3.2-4.6) | 107 | 3.2 (2.7-3.9) |
| Club foot – talipes equinovarus | 231 | 18 | 9 | 53 | 311 | 9.4 (8.4-10.6) | 280 | 8.5 (7.5-9.6) |
| Hip dislocation and/or dysplasia | 117 | 0 | 0 | 0 | 117 | 3.6 (2.9-4.3) | 112 | 3.4 (2.8-4.1) |
| Polydactyly | 125 | 3 | 0 | 8 | 136 | 4.1 (3.5-4.9) | 116 | 3.5 (2.9-4.2) |
| Syndactyly | 52 | 2 | 0 | 6 | 60 | 1.8 (1.4-2.3) | 48 | 1.5 (1.1-1.9) |
| **Other anomalies/syndromes** | | | | | | | | |
| Skeletal dysplasias | 24 | 3 | 0 | 34 | 61 | 1.9 (1.4-2.4) | z | z |
| Genetic syndromes + microdeletions | 104 | 5 | 1 | 22 | 132 | 4.0 (3.4-4.8) | z | z |
| **Chromosomal** | **735** | **58** | **27** | **824** | **1,644** | **49.9 (47.5-52.4)** | **z** | **z** |
| Down's syndrome | 394 | 23 | 8 | 398 | 823 | 25.0 (23.3-26.8) | z | z |
| Patau's syndrome | 7 | 2 | 1 | 63 | 73 | 2.2 (1.7-2.8) | z | z |
| Edwards' syndrome | 33 | 17 | 7 | 184 | 241 | 7.3 (6.4-8.3) | z | z |
| Turner's syndrome | 16 | 2 | 6 | 64 | 88 | 2.7 (2.1-3.3) | z | z |
| Klinefelter's syndrome | 14 | 0 | 0 | 3 | 17 | 0.5 (0.3-0.8) | z | z |

1 = Some of the cases shown in this table will have more than one anomaly and appear in more than one row of the table, so may not have had the TOPFA for the anomaly shown. Please see Glossary & Technical details document for more information about definitions and inclusion criteria
2 = Genetic conditions include those cases with an identified chromosomal anomaly, skeletal dysplasia, genetic syndrome and/or microdeletion
3 = 24+ weeks' gestation
4 = 20-23 weeks' gestation
5 = Termination of pregnancy with fetal anomaly
6 = This includes the following congenital heart anomalies: 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
7 = Congenital heart defects
8 = Gestational age
z = Not applicable

# Table 2: Birth prevalence (per 10,000 total births) and 95% confidence intervals by NCARDRS region according to congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| | East Midlands & South Yorkshire | Northern | South West | Thames Valley | Wessex | West Midlands | Yorkshire & Humber (YH) | All NCARDRS reporting regions | All NCARDRS reporting regions excluding YH | EUROCAT registries excluding NCARDRS reporting regions |
|---|---|---|---|---|---|---|---|---|---|---|
| **Total births** | **73,269** | **31,871** | **49,004** | **29,480** | **29,894** | **71,367** | **44,416** | **329,301** | **284,885** | **294,488** |
| **Total number of cases** | **1,458** | **713** | **1,074** | **634** | **627** | **1,446** | **800** | **6,752** | **5,952** | **7,987** |
| **Birth prevalence per 10,000 total births (95% CI)** | | | | | | | | | | |
| **All cases[1]** | **199.0 (188.9-209.5)** | **223.7 (207.6-240.7)** | **219.2 (206.3-232.7)** | **215.1 (198.7-232.5)** | **209.7 (193.7-226.8)** | **202.6 (192.3-213.3)** | **180.1 (167.9-193.0)** | **205.0 (200.2-210.0)** | **208.9 (203.7-214.3)** | **271.2 (265.3-277.2)** |
| Nervous system | 22.2 (19.0-25.9) | 22.3 (17.4-28.1) | 24.3 (20.1-29.1) | 23.1 (17.9-29.2) | 20.7 (15.9-26.6) | 27.6 (23.9-31.7) | 21.8 (17.7-26.6) | 23.6 (22.0-25.3) | 23.9 (22.1-25.7) | 24.9 (23.1-26.7) |
| Congenital heart defects | 63.3 (57.7-69.4) | 71.2 (62.3-81.1) | 59.0 (52.4-66.2) | 63.8 (55.0-73.6) | 61.6 (53.0-71.1) | 80.0 (73.6-86.8) | 55.4 (48.7-62.8) | 65.9 (63.1-68.7) | 67.5 (64.5-70.6) | 84.8 (81.5-88.2) |
| Severe CHD[2,3] | 26.2 (22.6-30.2) | 30.4 (24.7-37.1) | 23.9 (19.7-28.6) | 23.7 (18.5-30.0) | 26.8 (21.2-33.3) | 28.7 (24.9-32.9) | 23.4 (19.1-28.4) | 26.3 (24.5-28.1) | 26.7 (24.8-28.7) | 26.0 (24.2-27.9) |
| Respiratory | 2.9 (1.8-4.4) | 6.9 (4.3-10.5) | 7.1 (5.0-9.9) | 3.1 (1.4-5.8) | 5.7 (3.3-9.1) | 4.2 (2.8-6.0) | 5.4 (3.5-8.0) | 4.8 (4.1-5.6) | 4.7 (3.9-5.6) | 4.2 (3.5-5.1) |
| Oro-facial clefts | 16.2 (13.5-19.4) | 18.5 (14.1-23.9) | 13.9 (10.8-17.6) | 15.9 (11.7-21.2) | 17.7 (13.3-23.2) | 14.2 (11.5-17.2) | 14.4 (11.1-18.4) | 15.5 (14.2-16.9) | 15.7 (14.3-17.2) | 14.1 (12.8-15.6) |
| Digestive system | 18.2 (15.2-21.5) | 19.5 (14.9-24.9) | 19.6 (15.9-23.9) | 13.2 (9.4-18.1) | 22.7 (17.7-28.8) | 16.5 (13.7-19.8) | 18.9 (15.1-23.4) | 18.2 (16.8-19.7) | 18.1 (16.6-19.7) | 18.0 (16.5-19.6) |
| Abdominal wall defects | 8.9 (6.8-11.3) | 6.9 (4.3-10.5) | 11.2 (8.5-14.6) | 6.4 (3.9-10.1) | 6.7 (4.1-10.3) | 8.7 (6.7-11.1) | 5.4 (3.5-8.0) | 8.1 (7.2-9.1) | 8.5 (7.5-9.7) | 6.5 (5.6-7.5) |
| Urinary | 15.3 (12.6-18.4) | 27.6 (22.1-34.0) | 33.5 (28.5-39.0) | 26.5 (20.9-33.0) | 25.1 (19.7-31.4) | 16.7 (13.8-20.0) | 21.2 (17.1-25.9) | 22.2 (20.6-23.8) | 22.3 (20.6-24.1) | 39.6 (37.4-41.9) |
| Bilateral renal agenesis including Potter syndrome | 0.3 (0.0-1.0) | 1.9 (0.7-4.1) | 2.0 (1.0-3.8) | 2.4 (0.9-4.9) | 1.7 (0.5-3.9) | 1.7 (0.9-2.9) | 0.9 (0.2-2.3) | 1.4 (1.0-1.9) | 1.5 (1.1-2.0) | 1.5 (1.1-2.0) |
| Genital | 11.5 (9.1-14.2) | 16.9 (12.7-22.1) | 12.2 (9.3-15.8) | 9.8 (6.6-14.1) | 12.4 (8.7-17.1) | 12.3 (9.9-15.2) | 11.5 (8.6-15.1) | 12.2 (11.1-13.5) | 12.4 (11.1-13.7) | 24.1 (22.3-25.9) |

| | East Midlands & South Yorkshire | Northern | South West | Thames Valley | Wessex | West Midlands | Yorkshire & Humber (YH) | All NCARDRS reporting regions | All NCARDRS reporting regions excluding YH | EUROCAT registries excluding NCARDRS reporting regions |
|---|---|---|---|---|---|---|---|---|---|---|
| Limb | 18.8 (15.8-22.3) | 23.5 (18.5-29.5) | 33.9 (28.9-39.4) | 33.2 (27.0-40.5) | 24.1 (18.8-30.3) | 23.0 (19.6-26.8) | 20.5 (16.5-25.2) | 24.4 (22.8-26.2) | 25.0 (23.2-26.9) | 48.1 (45.6-50.6) |
| Limb reduction defects | 3.8 (2.5-5.5) | 2.8 (1.3-5.4) | 6.1 (4.1-8.7) | 3.1 (1.4-5.8) | 3.7 (1.8-6.6) | 3.4 (2.2-5.0) | 3.4 (1.9-5.6) | 3.8 (3.2-4.6) | 3.9 (3.2-4.7) | 5.1 (4.3-6.0) |
| Skeletal dysplasias | 2.3 (1.4-3.7) | 1.9 (0.7-4.1) | 2.4 (1.3-4.3) | 1.4 (0.4-3.5) | 1.7 (0.5-3.9) | 1.1 (0.5-2.2) | 2.0 (0.9-3.9) | 1.9 (1.4-2.4) | 1.8 (1.4-2.4) | 2.1 (1.6-2.7) |
| Genetic syndromes + microdeletions | 2.2 (1.2-3.5) | 6.3 (3.8-9.7) | 5.1 (3.3-7.5) | 2.4 (0.9-4.9) | 5.0 (2.8-8.3) | 4.8 (3.3-6.7) | 3.4 (1.9-5.6) | 4.0 (3.4-4.8) | 4.1 (3.4-4.9) | 5.4 (4.6-6.3) |
| Chromosomal | 46.1 (41.3-51.3) | 47.1 (39.8-55.2) | 57.3 (50.8-64.5) | 53.9 (45.9-63.0) | 55.9 (47.7-65.0) | 50.4 (45.4-55.9) | 42.6 (36.7-49.1) | 49.9 (47.5-52.4) | 51.1 (48.5-53.8) | 43.5 (41.2-46.0) |

1 = Some of the cases shown in this table will have more than one anomaly and appear in more than one row of the table
2 = This includes the following congenital heart anomalies: 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
3 = Congenital heart defects

## Table 3: Number of live births, live birth prevalence (per 10,000 total births) and 95% confidence intervals according to major congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | Number | Live birth prevalence per 10,000 live births (95% CI) |
|---|---|---|
| **All cases** | **4,867** | **148.4 (144.3-152.7)** |
| Nervous system | 299 | 9.1 (8.1-10.2) |
| Congenital heart defects | 1,812 | 55.3 (52.7-57.9) |
| Respiratory | 111 | 3.4 (2.8-4.1) |
| Oro-facial clefts | 456 | 13.9 (12.7-15.2) |
| Digestive system | 474 | 14.5 (13.2-15.8) |
| Abdominal wall defects | 158 | 4.8 (4.1-5.6) |
| Urinary | 565 | 17.2 (15.8-18.7) |
| Genital | 374 | 11.4 (10.3-12.6) |
| Limb | 633 | 19.3 (17.8-20.9) |
| Skeletal dysplasias | 24 | 0.7 (0.5-1.1) |
| Genetic syndromes + microdeletions | 104 | 3.2 (2.6-3.8) |
| Chromosomal | 735 | 22.4 (20.8-24.1) |

1 = Some of the cases shown in this table will have more than one anomaly and appear in more than 1 row of the table

## Table 4: Estimated numbers of cases of congenital anomalies in England based on the prevalence in the 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | Birth prevalence per 10,000 total births (95% CI) | Estimated number of cases in England | Birth prevalence (excluding YH[2]) per 10,000 total birth (95% CI) | Estimated number of cases in England |
|---|---|---|---|---|
| **Total births** | **z** | **666,050** | **z** | **666,050** |
| **All cases** | **205.0 (200.2-210.0)** | **13,657** | **208.9 (203.7-214.3)** | **13,916** |
| Nervous system | 23.6 (22.0-25.3) | 1,572 | 23.9 (22.1-25.7) | 1,590 |
| Congenital heart defects | 65.9 (63.1-68.7) | 4,387 | 67.5 (64.5-70.6) | 4,496 |
| Respiratory | 4.8 (4.1-5.6) | 320 | 4.7 (3.9-5.6) | 313 |
| Oro-facial clefts | 15.5 (14.2-16.9) | 1,034 | 15.7 (14.3-17.2) | 1,045 |
| Digestive system | 18.2 (16.8-19.7) | 1,214 | 18.1 (16.6-19.7) | 1,206 |
| Abdominal wall defects | 8.1 (7.2-9.1) | 540 | 8.5 (7.5-9.7) | 568 |
| Urinary | 22.2 (20.6-23.8) | 1,477 | 22.3 (20.6-24.1) | 1,487 |
| Genital | 12.2 (11.1-13.5) | 815 | 12.4 (11.1-13.7) | 823 |
| Limb | 24.4 (22.8-26.2) | 1,626 | 25.0 (23.2-26.9) | 1,667 |
| Skeletal dysplasias | 1.9 (1.4-2.4) | 123 | 1.8 (1.4-2.4) | 122 |
| Genetic syndromes + microdeletions | 4.0 (3.4-4.8) | 267 | 4.1 (3.4-4.9) | 274 |
| Chromosomal | 49.9 (47.5-52.4) | 3,325 | 51.1 (48.5-53.8) | 3,402 |

1 = Some of the cases shown in this table will have more than one anomaly and appear in more than one row of the table
2 = Yorkshire and Humber
z = Not applicable

## Table 5: Number, percentage and 95% confidence intervals for timing of first diagnosis and pregnancy outcome for all cases in 7 NCARDRS reporting regions, 2016

| Time of diagnosis | Pregnancy outcome | Number | Percentage (95% CI) |
|---|---|---|---|
| **All cases n=6,752 (100%)** | | **6,752** | **z** |
| **Antenatal n=4,159 (61.6%)** | **Total** | **4,159** | **100.0** |
| | TOPFA[1] | 1,608 | 38.7 (37.2-40.2) |
| | Miscarriage (20-23 weeks) | 37 | 0.9 (0.6-1.2) |
| | Stillbirth (24+ weeks) | 119 | 2.9 (2.4-3.4) |
| | Live birth | 2,395 | 57.6 (56.1-59.1) |
| **Postnatal n=2,249 (33.3%)** | **Total** | **2,249** | **100.0** |
| | TOP for other medical reason | 9 | 0.4 (0.2-0.8) |
| | Miscarriage (20-23 weeks) | 36 | 1.6 (1.1-2.2) |
| | Stillbirth (24+ weeks) | 68 | 3.0 (2.4-3.8) |
| | Live birth | 2,136 | 95.0 (94.0-95.8) |
| **Timing of diagnosis not known n=344 (5.1%)** | **Total** | **344** | **100.0** |
| | Miscarriage (20-23 weeks) | 4 | 1.2 (0.5-3.0) |
| | Stillbirth (24+ weeks) | 4 | 1.2 (0.5-3.0) |
| | Live birth | 336 | 97.7 (95.5-98.8) |

1 = Termination of pregnancy with fetal anomaly
z = Not applicable

## Table 6: Number, percentage and 95% confidence intervals for timing of diagnosis according to major congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | Total number of cases | Number antenatally diagnosed | Percentage (95% CI) |
|---|---|---|---|
| Nervous system | 777 | 622 | 80.1 (77.1-82.7) |
| Congenital heart defects | 2,169 | 815 | 37.6 (35.6-39.6) |
| Respiratory | 158 | 71 | 44.9 (37.4-52.7) |
| Oro-facial clefts | 511 | 270 | 52.8 (48.5-57.1) |
| Digestive system | 600 | 186 | 31.0 (27.4-34.8) |
| Abdominal wall defects | 267 | 233 | 87.3 (82.7-90.7) |
| Urinary | 730 | 543 | 74.4 (71.1-77.4) |
| Genital | 403 | 14 | 3.5 (2.1-5.7) |
| Limb | 804 | 309 | 38.4 (35.1-41.8) |
| Skeletal dysplasias | 61 | 42 | 68.9 (56.4-79.1) |
| Genetic syndromes + microdeletions | 132 | 41 | 31.1 (23.8-39.4) |
| Chromosomal | 1,644 | 870 | 52.9 (50.5-55.3) |

1 = Some of the cases shown in this table will have more than one anomaly and appear in more than 1 row of the table

## Table 7: Prevalence (per 10,000 total births) and 95% confidence intervals for terminations of pregnancy with fetal anomaly according to major congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | TOPFA[2] <20 weeks rate per 10,000 total births (95% CI) | TOPFA[2] 20+ weeks rate per 10,000 total births (95% CI) | Total TOPFA[2] rate per 10,000 total births (95% CI) |
|---|---|---|---|
| **All cases** | **28.1 (26.3-30.0)** | **17.4 (16.0-18.9)** | **49.1 (46.7-51.6)** |
| Nervous system | 6.5 (5.7-7.4) | 6.2 (5.4-7.1) | 13.5 (12.3-14.9) |
| Congenital heart defects | 2.6 (2.1-3.2) | 5.7 (4.9-6.6) | 8.5 (7.5-9.5) |
| Respiratory | 0.4 (0.2-0.6) | 0.6 (0.4-1.0) | 1.0 (0.7-1.4) |
| Oro-facial clefts | 0.5 (0.3-0.8) | 1.0 (0.7-1.4) | 1.5 (1.1-1.9) |
| Digestive system | 1.4 (1.0-1.9) | 1.5 (1.1-2.0) | 3.0 (2.5-3.7) |
| Abdominal wall defects | 2.4 (1.9-3.0) | 0.4 (0.2-0.6) | 2.9 (2.3-3.5) |
| Urinary | 1.7 (1.3-2.2) | 2.4 (1.9-3.0) | 4.3 (3.6-5.0) |
| Genital | 0.4 (0.2-0.6) | 0.2 (0.1-0.5) | 0.7 (0.4-1.0) |
| Limb | 1.7 (1.3-2.2) | 1.9 (1.4-2.4) | 3.8 (3.1-4.5) |
| Skeletal dysplasias | 0.4 (0.2-0.6) | 0.6 (0.4-1.0) | 1.0 (0.7-1.4) |
| Genetic syndromes + microdeletions | 0.3 (0.2-0.6) | 0.3 (0.2-0.6) | 0.7 (0.4-1.0) |
| Chromosomal | 17.9 (16.5-19.5) | 4.7 (4.0-5.5) | 25.0 (23.3-26.8) |

1 = Some of the cases shown in this table will have more than 1 anomaly and appear in more than 1 row of the table, so may not have had the TOPFA for the anomaly shown
2 = Termination of pregnancy with fetal anomaly

## Table 8: Number, percentage and 95% confidence intervals for type of termination of pregnancy with fetal anomaly for all cases in 7 NCARDRS reporting regions, 2016

| Type of TOPFA[1] | Number | Percentage (95% CI) |
|---|---|---|
| **All cases** | **1,617** | **100.0** |
| Feticide | 216 | 13.4 (11.8-15.1) |
| Medical TOPFA[1] | 795 | 49.2 (46.7-51.6) |
| Surgical TOPFA[1] | 125 | 7.7 (6.5-9.1) |
| Other/unknown method | 481 | 29.7 (27.6-32.0) |

1 = Termination of pregnancy with fetal anomaly

## Table 9: Numbers, mortality rates (per 10,000 births) and 95% confidence intervals for perinatal and infant deaths according to congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly[1] | Number of perinatal[2] deaths | Perinatal[2] mortality rate (per 10,000 total births) (95% CI) | Number of infant[3] deaths | Infant[3] mortality rate (per 10,000 live births) (95% CI) |
|---|---|---|---|---|
| **All anomalies** | 355 | 10.8 (9.7-12.0) | 336 | 10.2 (9.2-11.4) |
| Nervous system | 54 | 1.6 (1.2-2.1) | 51 | 1.6 (1.2-2.0) |
| Congenital heart defects | 107 | 3.2 (2.7-3.9) | 159 | 4.8 (4.1-5.7) |
| Respiratory | 16 | 0.5 (0.3-0.8) | 19 | 0.6 (0.3-0.9) |
| Oro-facial clefts | 14 | 0.4 (0.2-0.7) | 13 | 0.4 (0.2-0.7) |
| Digestive system | 55 | 1.7 (1.3-2.2) | 60 | 1.8 (1.4-2.4) |
| Abdominal wall defects | 22 | 0.7 (0.4-1.0) | 16 | 0.5 (0.3-0.8) |
| Urinary | 54 | 1.6 (1.2-2.1) | 45 | 1.4 (1.0-1.8) |
| Genital | 15 | 0.5 (0.3-0.8) | 18 | 0.5 (0.3-0.9) |
| Limb | 53 | 1.6 (1.2-2.1) | 40 | 1.2 (0.9-1.7) |
| Skeletal dysplasias | 7 | 0.2 (0.1-0.4) | 5 | 0.2 (0.0-0.4) |
| Genetic syndromes + microdeletions | 10 | 0.3 (0.1-0.6) | 18 | 0.5 (0.3-0.9) |
| Chromosomal | 89 | 2.7 (2.2-3.3) | 83 | 2.5 (2.0-3.1) |

1 = Some of the cases shown in this table will have more than 1 anomaly and appear in more than 1 row of the table. Caution should be taken when interpreting these data as some cases may have died from 1 anomaly but will also appear in the data for the other (possibly less severe) anomalies.
2 = Stillbirths and early neonatal deaths (24+ weeks' gestation to 6 days old)
3 = Early, late and post neonatal deaths (less than 1 year old)

## Table 10: Birth prevalence (per 10,000 total births) and 95% confidence intervals according to maternal age for each congenital anomaly subgroup in 7 NCARDRS reporting regions, 2016

| Congenital anomaly | Total | Under 20 | 20-24 | 25-29 | 30-34 | 35-39 | 40 and over |
|---|---|---|---|---|---|---|---|
| **Total births** | **329,301** | **12,051** | **53,747** | **97,086** | **100,795** | **53,461** | **12,161** |
| **Total number of cases** | **6,752[a]** | **266** | **1,062** | **1,717** | **1,846** | **1,325** | **525** |
| **Birth prevalence per 10,000 total births (95% CI)** | | | | | | | |
| **All cases** | **205.0 (200.2-210.0)** | **220.7 (195.0-248.9)** | **197.6 (185.9-209.8)** | **176.9 (168.6-185.4)** | **183.1 (174.9-191.7)** | **247.8 (234.7-261.6)** | **431.7 (395.6-470.2)** |
| **Non-genetic anomalies[1]** | **150.9 (146.7-155.1)** | **185.0 (161.6-211.0)** | **166.3 (155.6-177.6)** | **144.6 (137.2-152.4)** | **141.1 (133.8-148.6)** | **154.5 (144.2-165.4)** | **160.3 (138.6-184.5)** |
| Nervous system | 21.0 (19.4-22.6) | 29.0 (20.2-40.4) | 24.9 (20.9-29.5) | 19.8 (17.1-22.8) | 19.9 (17.3-22.9) | 19.6 (16.1-23.8) | 18.1 (11.3-27.4) |
| Congenital heart defects | 52.6 (50.1-55.1) | 49.0 (37.3-63.1) | 52.7 (46.7-59.2) | 51.1 (46.7-55.8) | 50.7 (46.4-55.3) | 58.7 (52.4-65.6) | 55.9 (43.4-70.9) |
| Respiratory | 4.2 (3.5-5.0) | 4.1 (1.3-9.7) | 5.2 (3.5-7.5) | 4.0 (2.9-5.5) | 3.9 (2.8-5.3) | 4.5 (2.9-6.7) | 3.3 (0.9-8.5) |
| Oro-facial clefts | 13.8 (12.5-15.1) | 15.8 (9.5-24.6) | 13.8 (10.8-17.3) | 13.4 (11.2-15.9) | 13.2 (11.0-15.6) | 14.8 (11.7-18.4) | 14.8 (8.8-23.4) |
| Digestive system | 15.1 (13.8-16.5) | 14.1 (8.2-22.6) | 17.9 (14.5-21.8) | 15.7 (13.3-18.4) | 13.2 (11.0-15.6) | 15.0 (11.9-18.6) | 15.6 (9.4-24.4) |
| Abdominal wall defects | 6.6 (5.8-7.6) | 25.7 (17.5-36.5) | 13.2 (10.3-16.7) | 5.9 (4.4-7.6) | 3.6 (2.5-4.9) | 2.4 (1.3-4.2) | 8.2 (3.9-15.1) |
| Urinary | 19.8 (18.3-21.4) | 24.9 (16.8-35.5) | 19.9 (16.3-24.1) | 19.6 (16.9-22.6) | 18.8 (16.2-21.6) | 19.3 (15.7-23.4) | 27.1 (18.7-38.1) |
| Genital | 11.2 (10.1-12.4) | 10.8 (5.7-18.5) | 10.6 (8.0-13.7) | 10.6 (8.7-12.9) | 10.1 (8.3-12.3) | 13.7 (10.7-17.2) | 17.3 (10.7-26.4) |
| Limb | 21.2 (19.6-22.8) | 29.9 (20.9-41.4) | 22.9 (19.0-27.3) | 19.8 (17.1-22.8) | 20.6 (17.9-23.6) | 20.6 (16.9-24.8) | 23.0 (15.3-33.3) |
| **Genetic anomalies** | **54.2 (51.7-56.7)** | **35.7 (25.8-48.1)** | **31.3 (26.7-36.4)** | **32.2 (28.8-36.0)** | **42.1 (38.2-46.3)** | **93.3 (85.3-101.9)** | **271.4 (242.9-302.3)** |
| Skeletal dysplasias | 1.9 (1.4-2.4) | 0.8 (0.0-4.7) | 1.5 (0.6-2.9) | 1.1 (0.6-2.0) | 2.7 (1.8-3.9) | 1.7 (0.8-3.2) | 4.1 (1.3-9.6) |
| Genetic syndromes and microdeletions | 4.0 (3.4-4.8) | 5.0 (1.8-10.9) | 4.8 (3.2-7.1) | 3.5 (2.4-4.9) | 4.0 (2.8-5.4) | 3.7 (2.3-5.8) | 4.9 (1.8-10.8) |
| Chromosomal | 49.9 (47.5-52.4) | 29.9 (20.9-41.4) | 26.4 (22.3-31.1) | 28.9 (25.7-32.5) | 37.4 (33.7-41.4) | 89.6 (81.8-98.0) | 264.8 (236.7-295.3) |

a = Includes age not known (0.2% of all cases)
1 = Includes those cases with 1 or more congenital anomaly with no identified anomalies that are chromosomal, skeletal dysplasias, genetic syndromes or microdeletions

## Table 11: Eleven auditable structural anomalies with FASP detection rates and early detection in 46 English reporting trusts, 2015 to 2016

| Anomaly | Total cases[1] | FASP exclusions: total[2] | FASP exclusions: early detections | All cases screened in FASP window | | FASP detection rate % (95% CI) | Detection rate including early detections % (95% CI) | FASP target % |
|---|---|---|---|---|---|---|---|---|
| | | | | detected (screen +ve) | undetected (screen –ve) | | | |
| Anencephaly | 132 | 127 | 119 | 5 | 0 | 100.0 (56.6-100.0) | 100.0 (97.0-100.0) | 98 |
| Spina bifida | 134 | 34 | 28 | 93 | 7 | 93.0 (86.3-96.6) | 94.5 (89.1-97.3) | 90 |
| Cleft lip +/- palate | 242 | 47 | 15 | 155 | 39 | 79.5 (73.3-84.6) | 81.0 (75.1-85.7) | 75 |
| Diaphragmatic hernia | 73 | 10 | 5 | 38 | 25 | 60.3 (48.0-71.5) | 63.2 (51.4-73.7) | 60 |
| Gastroschisis | 91 | 75 | 71 | 15 | 1 | 93.8 (71.7-98.9) | 98.9 (93.8-99.8) | 98 |
| Exomphalos | 119 | 110 | 99 | 6 | 3 | 66.7 (35.4-87.9) | 97.2 (92.1-99.1) | 80 |
| Serious cardiac | 365 | 62 | 41 | 195 | 107 | 64.4 (58.8-69.5) | 68.6 (63.5-73.3) | 50 |
| Transposition of the great arteries (TGA) | 84 | 10 | 3 | 46 | 28 | 62.2 (50.8-72.4) | 63.6 (52.5-73.5) | 50 |
| Tetralogy of Fallot (ToF) | 86 | 13 | 9 | 42 | 30 | 57.5 (46.1-68.2) | 62.2 (51.4-71.9) | 50 |
| Atriocentricular septal defect (AVSD) | 133 | 31 | 21 | 59 | 43 | 57.8 (48.1-67.0) | 65.0 (56.3-72.9) | 50 |
| Hypoplastic left heart (HLH) | 62 | 8 | 8 | 48 | 6 | 88.9 (77.8-94.8) | 90.3 (80.5-95.5) | 50 |
| Bilateral renal agenesis | 32 | 8 | 4 | 17 | 7 | 70.8 (50.8-85.1) | 75.0 (56.6-87.3) | 85 |
| Lethal skeletal dysplasia | 23 | 8 | 7 | 10 | 5 | 66.7 (41.7-84.8) | 77.3 (56.6-89.9) | 60 |
| Edwards' syndrome | 185 | 135 | 90 | 36 | 14 | 72.0 (58.3-82.5) | 90.0 (83.9-93.9) | 95 |
| Patau's syndrome | 80 | 60 | 31 | 17 | 3 | 85.0 (64.0-94.8) | 94.1 (84.1-98.0) | 95 |

1 = Total complete cases reported to NCARDRS
2 = Ineligible (late/no booking, early fetal loss/TOP before screening), declined, early detections