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Population-based birth defects data in the United States, 2011-2015: A focus on eye and ear defects

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DISCLAIMER

The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

Abstract

Background/Objectives: In this data brief, we examine major eye and ear anomalies (anophthalmia/microphthalmia, anotia/microtia, and congenital cataract) for a recent 5-year birth cohort using data from 30 population-based birth defects surveillance programs in the United States.

Methods: As a special call for data for the 2018 NBDPN Annual Report, state programs reported expanded data on eye/ear anomalies for birth years 2011–2015. We calculated the combined overall prevalence (per 10,000 live births) and 95% confidence intervals (CI), for the three anomalies as well as by maternal age, maternal race/ethnicity, infant sex, laterality, presence/absence of other major birth defects, and case ascertainment methodology utilized by the program (active vs. passive).

Results: The overall prevalence estimate (per 10,000 live births) was 1.5 (95% CI: 1.4–1.5) for anophthalmia/microphthalmia, 1.5 (95% CI: 1.4–1.6) for congenital cataract, and 1.8 (95% CI: 1.7–1.8) for anotia/microtia. Congenital cataract prevalence varied little by maternal race/ethnicity, infant sex, or case ascertainment methodology; prevalence differences were more apparent across strata for anophthalmia/microphthalmia and anotia/microtia. Prevalence among active vs. passive ascertainment programs was 50% higher for anophthalmia/microphthalmia (1.9 vs. 1.2) and two-fold higher for anotia/microtia (2.6 vs. 1.2). Anophthalmia/microphthalmia was more likely than other conditions to co-occur with other birth defects. All conditions were more frequent among older mothers (40+ years).

Conclusions: This data brief provides recent prevalence estimates for anophthalmia/microphthalmia, congenital cataract, and anotia/microtia that address a data gap by examining pooled data from 30 population-based surveillance systems, covering a five-year birth cohort of about 12.4 million births.

Keywords

anophthalmia; anotia; birth defects; cataract; congenital; microphthalmia; microtia; population-based surveillance

1 | INTRODUCTION

The National Birth Defects Prevention Network (NBDPN) has collaborated with population-based birth defects surveillance programs in the United States and the Centers for Disease Control and Prevention (CDC) annually since 2000 to publish state-level data on major structural birth defects as part of the Congenital Malformations Surveillance Report. The NBDPN updated the list of major birth defects in 2014 after a thorough review of conditions. Defects included on the list are those with high public health importance, those that are generally diagnosed within the first year of life, those amenable to potential prevention/intervention strategies, and those that can be monitored as a separate condition (Mai et al., 2014).

The 2018 NBDPN Congenital Malformations Surveillance Report includes state-specific birth defects counts and prevalence estimates from 41 population-based birth defects

surveillance systems, reporting up to 47 major birth defects covering the central nervous system, eye and ear, cardiovascular system, gastrointestinal system, genitourinary system, musculoskeletal system, as well as chromosomal conditions, described in Table 1. A program directory is included to describe individual population-based birth defects programs, available on pages [S123-S177]. The directory covers each program's case definition, surveillance methodology, sources of birth defects information, data collection procedures, and program contacts. Finally, the NBDPN report features a data brief that pools data from 30 surveillance programs to provide a more detailed description of eye and ear anomalies.

2 | STATE-SPECIFIC DATA COLLECTION AND PRESENTATION OF 47 MAJOR BIRTH DEFECTS

2.1 | Data collection

The NBDPN Data Committee, in collaboration with the CDC, issued a call for data to state and territorial birth defects surveillance programs to report birth defects cases for a birth cohort beginning with January 1, 2011 and end-ing with December 31, 2015. The call for data included instructions and a data dictionary for each of the major birth defects on the NBDPN list (Table 1) by year of birth, maternal race/ethnicity, maternal age at delivery, and infant sex. Maternal race/ethnicity was stratified by U.S. Census group: non-Hispanic white, non-Hispanic black, Hispanic, non-Hispanic Asian/Pacific Islander, non-Hispanic American Indian/Alaska Native, and other/unknown. Maternal age at delivery was stratified by the following age groups: < 20, 20–24, 25–29, 30–34, 35–39, ≥40 years of age, and unknown. Infant sex was collected as male, female, and unknown.

A sample SAS file and an Excel template were provided to state programs to aid with data submission. In addition, a PDF fillable directory with prepopulated responses about each program (from their previous submission) was provided for program review and updates. Following data and directory profile submission to CDC for central cleaning and processing using SAS 9.4 (Cary, NC), CDC staff worked with state programs to review and validate the submitted information. Review and approval by state birth defects programs were completed prior to final creation of data tables and directory profiles.

2.2 | Data presentation

A total of 41 state and territorial birth defects surveillance programs submitted data for this NBDPN Data Report. Data for the birth cohort 2011–2015 are presented in two separate tables for each state/territorial surveillance program (Supporting Information). The first table for each surveillance program displays counts and prevalence (per 10,000 live births) by five maternal racial/ethnic groups. The second table presents counts and prevalence by two maternal age categories (<35 years, ≥35 years) for gastroschisis and trisomies 13, 18, and 21. Maternal age groups were collapsed from six categories into two larger age groups to ensure adequate cell counts. The prev-alence for all defects (excluding congenital posterior

SUPPORTING INFORMATION
Additional supporting information may be found online in the Supporting Information section at the end of the article.

urethral valves [CPUV], hypospadias, and Turner syndrome) was calculated as the count of cases within each stratum of a maternal or child characteristic—regardless of pregnancy outcome (i.e., live birth, stillbirth, spontaneous/elective termination)— divided by the total number of live births within the same stratum (Mason, Kirby, Sever, & Langlois, 2005). It is presented as prevalence per 10,000 total live births. For CPUV and hypospadias, the prevalence is per 10,000 male live births; for Turner syndrome, it is per 10,000 female live births.

To address variations in surveillance methodology, footnotes accompany each table to describe differences in case inclusion criteria, data sources, coding system(s) used, and/or case ascertainment methodology. The program directory in the Supporting Information contains more in-depth information on each program's data collection methods and potential sources of variation.

3 | DATA BRIEF ON EYE AND EAR ANOMALIES

3.1 | Background

The featured data brief for the 2018 Data Report focus on eye and ear anomalies, specifically anophthalmia/micro-phthalmia, congenital cataract, and anotia/microtia for birth years 2011–2015. Studies estimating the prevalence of major eye and ear anomalies from large-scale population-based data are limited. This brief provides an opportunity to examine the prevalence of these major eye and ear anomalies from population-based surveillance systems across the United States for a recent birth cohort.

3.1.1 | Eye anomalies—Anophthalmia/microphthalmia and congenital cataract are common defects of the eye. Anophthalmia is the total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit, whereas microphthalmia is defined by a reduced volume of the eye (the corneal diameter is usually <10 mm, or the anteroposterior globe diameter is <20 mm) (NBDPN, 2015). Congenital cataract is an opacity of the lens of the eye that has its origin prenatally (NBDPN, 2015). It is a common cause of preventable blindness in children (Gilbert & Foster, 2001).

Reported prevalence of anophthalmia/microphthalmia ranges from 1.87 per 10,000 births (Parker et al., 2010) to 3.00 per 10,000 births (Chambers et al., 2018). Bhatti et al. (2003) reported the prevalence of congenital cataract to be 2.03 per 10,000 births. In a 2010 case-control study, Pra-kalapakorn, Rasmussen, Lambert, and Honein (2010) reported 59% of congenital cataract cases had bilateral cataracts and 41% unilateral.

Anophthalmia/microphthalmia appears to be associated with some maternal and infant factors. For instance, it has been reported that the birth prevalence of these defects increases with advanced maternal age (Kallen & Tornqvist, 2005; Shaw et al., 2005). In addition, mothers of infants born with anophthalmia/microphthalmia or congenital cataract are more likely to be of lower socioeconomic status, unmarried, unemployed, and have fewer years of education (i.e., <12 years) (Puho, Vogt, Csaky-Szunyogh, Metneki, & Czeizel, 2008; Shaw et al., 2005). The risk for these conditions, especially bilateral microphthalmia, increases with multifetal pregnancies and higher parity (Shaw et al., 2005).

The prevalence of these defects has also been observed to differ by infant sex. For instance, the prevalence of anophthalmia/microphthalmia is higher in females than in males (Forrester & Merz, 2006). Haargaard, Wohlfahrt, Rosenberg, Fledelius, and Melbye (2005) reported that cases of bilateral isolated cataract were predominately male while unilateral isolated cases were predominately female.

A majority of anophthalmia/microphthalmia cases are associated with other congenital malformations. These primarily include anomalies of the central nervous system, facial anomalies, cardiovascular anomalies, chromosomal abnormalities, and musculoskeletal/limb anomalies. Cham-bers et al. (2018) found more than half (55.7%) of cases of anophthalmia/microphthalmia also had a chromosomal abnormality or syndrome, most commonly trisomy 13 or 21. That study also reported that 92.4% of nonsyndromic cases (i.e., without a known chromosomal abnormality or genetic syndrome) had at least one additional co-occurring birth defect (Chambers et al., 2018). In contrast, Bhatti et al. (2003) found that about a quarter of congenital cataract cases (22%) occur as part of a syndrome—most commonly trisomy 13—and another 20% occurred in conjunction with other birth defects.

3.1.2 | Ear anomalies—Defects of the ear include anotia (total absence of the external ear and canal) and microtia (malformation or hypoplasia of the external ear) (NBDPN, 2015). The prevalence of these conditions is approximately 3.06 per 10,000 live births, with a higher prevalence of isolated cases compared to those that occur with other defects or as part of a syndrome (Deng et al., 2016). Unilateral microtia, especially right-sided microtia, is the most common form (Yamauchi et al., 2012). Compared to the eye anomalies evaluated in this data brief, there are substantial racial differences for anotia/microtia. Anotia/microtia occurs more frequently among infants born to Hispanic mothers, either U.S.- or foreign-born, as well as infants born to American Indian/Alaska Native, Filipino and Chinese mothers when compared to other racial/ethnic groups (Hoyt et al., 2014; Canfield et al., 2014; Canfield, Langlois, Nguyen, & Scheuerle, 2009). Similar to defects of the eye, the prevalence of anotia/microtia increases with higher maternal age and lower maternal education (Canfield et al., 2009; Deng et al., 2016). In addition, anotia/microtia is more common among males, especially in isolated forms (Yamauchi et al., 2012).

Anotia and microtia are also associated with other congenital malformations. The most common associations are found in the craniofacial region, cardiovascular system, and musculoskeletal system, with the proportion of cooccurrence for craniofacial and limb defects increasing as the severity of the microtia increases (Luquetti et al., 2013; Yamauchi et al., 2012). More specifically, microtia is often accompanied by congenital heart defects, cleft lip and/or palate, vertebral defects, and anomalies of the extremities. Multiple malformations among infants with anotia/microtia are associated with low birthweight and increased maternal age (Yamauchi et al., 2012).

Because of the combined prevalence and impact of these conditions and limited recent studies, we wanted to examine the differences in the birth prevalence of these conditions by key characteristics, such as maternal race/ethnicity, maternal age, and infant sex.

3.2 | Methods

As a special call for data for the 2018 NBDPN Annual Report, state programs were invited to report expanded data on eye and ear anomalies. The specific codes for anophthalmia/microphthalmia, congenital cataract, and anotia/microtia is provided in Table 1. Data requested included case-level information by year of birth, maternal race/ethnicity, maternal age at delivery, infant sex, laterality (either in specific CDC/BPA code or as a separate field), and co-occurring birth defects. Programs were asked to submit information on any co-occurring birth defects they collect, including major and minor defects; however, this report presents co-occurring congenital anomalies grouped by organ system, as specified by ICD-9-CM codes (740–759 - congenital anomalies) and ICD-10-CM codes (Q00-Q99—congenital malformations, deformations, and chromosomal abnormalities), and excludes any birth defects codes outside these ranges. The data submission process and review followed a similar approach to the main annual report data submission. CDC obtained a nonresearch determination for the project.

Analyses were performed using SAS 9.4 (Cary). The 95% confidence intervals were calculated using exact Poisson methodology for prevalence estimates and exact binomial for measures of percent (Daly, 1992). Prevalence estimates are reported as the number of cases per 10,000 live births. A second CDC analyst validated the results presented in the tables.

4 | RESULTS

A total of 30 state and territorial birth defects surveillance programs, covering a total of 12,441,847 births from 2011 to 2015, provided expanded data for eye and ear anomalies. Table 2 presents the counts and birth prevalence for eye and ear anomalies by case ascertainment methodology, maternal race/ethnicity, maternal age, and infant sex. Pooling data from all 30 participating surveillance programs, the prevalence estimates (per 10,000 live births) were: 1.5 for anophthalmia/microphthalmia (1,785 cases), 1.5 for congenital cataract (1,818 cases), and 1.8 for anotia/microtia (2,206 cases).

Prevalence estimates per 10,000 live births were consistent by case ascertainment methodology for congenital cataract, but differences were seen when comparing case ascertainment methodology for anophthalmia/microphthalmia (1.9 for active versus 1.2 for passive) and for anotia/ microtia (2.6 for active versus 1.2 for passive).

Variations in prevalence were observed among maternal racial/ethnic groups (Table 2), with infants born to Hispanic and non-Hispanic American Indian/Alaska Native mothers having the highest prevalence (per 10,000) of anophthalmia/microphthalmia (both 1.9) and infants born to Hispanic mothers having the highest prevalence of anotia/microtia (3.6). For anotia/microtia, infants born to non-Hispanic American Indian/Alaska Native (2.6) and non-Hispanic Asian/Pacific Islander (2.4) mothers also had higher prevalence than other maternal racial/ethnic groups while the prevalence among infants born to non-Hispanic black mothers was substantially lower than other groups (0.8). Conversely, little variation among maternal racial/ethnic group was observed for congenital cataract cases.

When evaluating differences in prevalence by maternal age for all three conditions, the highest estimates (per 10,000 live births) were observed among infants born to mothers who were 40+ years of age at the time of delivery (3.7 for anophthalmia/microphthalmia, 2.5 for congenital cataract, and 3.2 for anotia/microtia).

Prevalence estimates (per 10,000 live births) were similar by infant sex for congenital cataract, but anophthalmia/ microphthalmia was more prevalent in females compared to males (1.6 vs. 1.4, respectively) and anotia/microtia was more prevalent in males (1.9 vs. 1.6) (Table 2).

Thirteen of the 30 state and territorial surveillance programs contributing data to the data brief were able to report laterality information for these defects. Table 3 presents laterality (unilateral and bilateral)—with unilaterality further divided into left, right, and not specified—for the eye and ear anomalies. Cases of congenital cataract were similarly split between unilateral and bilateral, although 23.0% were reported to be unknown laterality or were reported as anterior polar (i.e., had no laterality assigned). For anophthalmia/ microphthalmia, 61.3% of cases were reported as bilateral compared to 38.7% unilateral cases, while the inverse was observed for anotia/microtia (79.0% unilateral cases versus 21.0% bilateral cases). The unilateral cases for anophthalmia/microphthalmia were similarly split between left and right, but for anotia/microtia more unilateral right cases (49.2%) were reported compared to unilateral left cases (29.5%).

Table 4 displays co-occurring birth defects by organ system for the eye and ear anomalies. Co-occurring birth defects were most commonly observed among children with anophthalmia/microphthalmia (74.1%), intermediate for anotia/microtia cases (53.6%) and least frequent among congenital cataract cases (35.9%). Cardiovascular defects represented the most common co-occurring group of birth defects for all three conditions (43.0% of anophthalmia/microphthalmia cases, 18.8% of congenital cataract cases, and 27.7% of anotia/microtia cases). A large proportion of anophthalmia/microphthalmia cases had co-occurring ear, face, and/or neck defects (34.2%), musculoskeletal and/or limb defects (25.2% musculoskeletal, 28.9% limbs, and 29.4% other musculoskeletal), and/or chromosomal conditions (29.1%). Anotia/microtia cases also showed a moderate proportion of co-occurring musculoskeletal and limb defects (14.2% musculoskeletal, 11.8% limbs, and 23.3% other musculoskeletal).

5 | DISCUSSION

This report provides recent population-based prevalence estimates for eye and ear anomalies for a cohort of about 12.4 million births from 30 surveillance systems in the United States. In addition, our analysis provides new data on cooccurrence with other congenital anomalies by organ system.

The prevalence estimates presented in this data brief for all three conditions are lower than some previously reported in the literature (Bhatti et al., 2003; Chambers et al., 2018; Deng et al., 2016; Parker et al., 2010) but estimates for anophthalmia/microphthalmia and anotia/microtia are higher than what was reported in the 2015 NBDPN Annual Report data brief

(Mai et al., 2015). The overall prevalence for congenital cataract we observed is similar to that found in the 2015 report; however, the 2015 report shows a difference in cataract prevalence by case ascertainment methodology while our new prevalence estimates do not show a similar difference (Mai et al., 2015). Variations can be expected given different cohort periods, but another contributing factor might be variations in how the conditions are ascertained. As anophthalmia/microphthalmia and anotia/microtia include a range of severity, a wider range of prevalence estimates can be observed compared to a condition with a narrow range of severity, such as congenital cataract, where the estimates appear to be more consistent across programs and birth cohort periods.

Our results show a slightly higher proportion of anotia/ microtia with co-occurring defects compared to isolated ano-tia/microtia. This is different from what was observed by Deng et al. (2016), who found a higher prevalence of isolated anotia/microtia. Our inclusion of the entire range of congenital structural malformations ICD-9-CM/ICD-10-CM codes, which likely include a more heterogeneous group of structural malformations, could be the explanation for this difference. We evaluated co-occurrence by organ system for active and passive case-finding programs separately and found a similar proportion of co-occurring defects for all three eye/ear anomalies regardless of case-ascertainment methodology (data not shown).

Our results are similar to previous analyses of anotia/microtia with respect to the unilateral versus bilateral and left-right unilateral presentation, as well as prevalence by infant sex (Yamauchi et al., 2012). However, while a case-control study by Prakalapakorn et al. (2010) found a higher proportion of bilateral congenital cataract cases compared to unilateral cases, our results show a more even division for bilateral and unilateral cases.

Consistent with previous reports, the vast majority of anophthalmia/microphthalmia cases co-occurred with other birth defects (74.1%). Some of the commonly affected organ systems include ear/face/neck, musculoskeletal system, central nervous system anomalies, and cardiovascular defects. However, variations were observed by degree of cooccurrence among the studies, most likely due to inclusion criteria. For example, our results showed a higher proportion of cardiac conditions than Chambers et al. (2018). This could be due to cardiac conditions occurring as part of chromosomal syndromes; however, we decided not to remove chromosomal abnormalities to be able to present a complete description of all cases.

This report has several limitations. Variations in case ascertainment methodology and anomalies collected by the different state/territorial programs could contribute to variations in the quantity and range of co-occurring conditions reported. Some programs provided a more comprehensive list of co-occurring conditions while other programs captured a narrower list of conditions. The birth defects we evaluated for co-occurrence were also limited to the congenital anomalies range of ICD-9-CM codes and the congenital malformations, deformations, and chromosomal abnormalities range of ICD-10-CM. As a result, conditions that potentially may be considered congenital anomalies outside these ranges are excluded. However, by restricting the code range, the co-occurring data presented are more consistent across programs.

Our results are limited to cases with known laterality. For congenital cataract, it is unclear how the high percentage of unknown or unspecified cases (including several anterior polar cases where laterality could not be assigned) may have affected the reported proportions of laterality for this condition.

Finally, some state/territorial surveillance programs report recent years (mainly 2015) of their surveillance data as provisional. In October 2015, the coding system transitioned from ICD-9-CM to ICD-10-CM. An evaluation is in process to determine which conditions might have been affected by the transition, but we expect minimal impact for this report as this affects only 5% of the birth cohort (three out of 60 months of data).

6 | CONCLUSION

The 2018 NBDPN Congenital Malformations Surveillance Report includes data from 41 state and territorial population-based birth defect surveillance programs, reporting up to 47 major birth defects. The report continues to provide important and updated information on the occurrence of major birth defects in the United States. This year's data brief on eye and ear anomalies, using pooled surveillance data from 30 states, provide population-based prevalence estimates along with data on co-occurrence with conditions by organ system for a cohort of about 12.4 million births. The updated prevalence estimates for anophthalmia/microphthalmia, congenital cataract, and anotia/microtia may provide a basis for future epidemiologic studies to better understand the impact of these conditions.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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TABLE 1

Birth defects for the National Birth Defects Prevention Network (NBDPN) annual report by disease classification codes

Birth defects	ICD-9-CM codes	ICD-10-CM codes	CDC/BPA codes
Central nervous system			
Anencephaly	740.0–740.1	Q00.0-Q00.1	740.00–740.10
Spina bifida without anencephaly	741.0, 741.9 w/o 740.0–740.1	Q05.0-Q05.9, Q07.01, Q07.03 w/o Q00.0-Q00.1	741.00–741.99 w/o 740.00–740.10
Encephalocele	742.0	Q01.0-Q01.9	742.00–742.09
Holoprosencephaly	742.2	Q04.2	742.26
Eye[a]			
Anophthalmia/microphthalmia	743.0, 743.1	Q11.0-Q11.2	743.00–743.10
Congenital cataract	743.30–743.34	Q12.0	743.32
Ear[a]			
Anotia/microtia	744.01, 744.23	Q16.0, Q17.2	744.01, 744.21
Cardiovascular			
Common truncus (TA)	745.0	Q20.0	745.00 (excluding 745.01)
Transposition of the great arteries (TGA)	745.10, 0.12, 0.19	Q20.3, Q20.5	745.10–745.12, 745.18–745.19
[b]Dextro-Transposition of the great arteries (d-TGA)	745.10	Q20.3	745.10, 745.11, 745.18, 745.19
Tetralogy of Fallot (TOF)	745.2	Q21.3	745.20–745.21, 747.31
Ventricular septal defect	745.4	Q21.0	745.40–745.49 (excluding 745.487, 745.498)
Atrial septal defect	745.5	Q21.1	745.51–745.59
Atrioventricular septal defect (endocardial cushion defect)	745.60, 0.61, 0.69	Q21.2	745.60–745.69, 745.487
Pulmonary valve atresia and stenosis	746.01, 746.02	Q22.0, Q22.1	746.00, 746.01
[b]Pulmonary valve atresia	746.01	Q22.0	746.00
Tricuspid valve atresia and stenosis	746.1	Q22.4	746.100, 746.106 (excluding 746.105)
[b]Tricuspid valve atresia	746.1	Q22.4	746.100
Ebstein anomaly	746.2	Q22.5	746.20
Aortic valve stenosis	746.3	Q23.0	746.30
Hypoplastic left heart syndrome	746.7	Q23.4	746.70
Coarctation of aorta	747.10	Q25.1	747.10–747.19
Total anomalous pulmonary venous connection (TAPVC)	747.41	Q26.2	747.42

Birth defects	ICD-9-CM codes	ICD-10-CM codes	CDC/BPA codes
Single ventricle	745.3	Q20.4	745.3
Interrupted aortic arch (IAA)	747.11	Q25.2, Q25.4	747.215–747.217, 747.285
Double outlet right ventricle (DORV)	745.11	Q20.1	745.13–745.15
Orofacial			
Cleft palate alone (without cleft lip)	749.0	Q35.1-Q35.9	749.00–749.09
Cleft lip alone (without cleft palate)	749.1	Q36.0-Q36.9	749.10–749.19
Cleft lip with cleft palate	749.20–749.25	Q37.0-Q37.9	749.20–749.29
Choanal atresia	748.0	Q30.0	748.00
Gastrointestinal			
Esophageal atresia/tracheoesophageal fistula	750.3	Q39.0-Q39.4	750.30–750.35
Rectal and large intestinal atresia/stenosis	751.2	Q42.0-Q42.9	751.20–751.24
Biliary atresia	751.61	Q44.2-Q44.3	751.65
Small intestinal atresia/stenosis	751.1	Q41.0-Q41.9	751.10–751.19
Genitourinary			
Renal agenesis/hypoplasia	753.0	Q60.0-Q60.6	753.00–753.01
Bladder exstrophy	753.5	Q64.10, Q64.19	753.50
Hypospadias	752.61	Q54.0-Q54.9 (excluding Q54.4)	752.60–752.62 (excluding 752.61 and 752.621)
Congenital posterior urethral valves	753.6	Q64.2	753.60
Cloacal exstrophy	751.5	Q64.12	751.555
Musculoskeletal			
Gastroschisis	756.73	Q79.3	756.71
Omphalocele	756.72	Q79.2	756.70
Diaphragmatic hernia	756.6	Q79.0, Q79.1	756.610–756.617
Limb deficiencies (reduction defects)	755.2–755.4	Q71.0-Q71.9, Q72.0-Q72.9, Q73.0-Q73.8	755.20–755.49
Craniosynostosis	No specific code	Q75.0	756.00–756.03
Clubfoot	754.51, 754.70	Q66.0, Q66.89	754.50, 754.73 (excluding 754.735)
Chromosomal			
Trisomy 13	758.1	Q91.4-Q91.7	758.10–758.19
Trisomy 21 (down syndrome)	758.0	Q90.0-Q90.9	758.00–758.09
Trisomy 18	758.2	Q91.0– 91.3	758.20–758.29
Turner syndrome	758.6	Q96.0-Q96.9	758.60–758.69

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Birth defects	ICD-9-CM codes	ICD-10-CM codes	CDC/BPA codes
Deletion 22 q11.2	758.32	Q93.81	758.37

ICD-9-CM: International Classification of Diseases, 9th Revision, Clinical Modification; ICD-10-CM: International Classification of Diseases, 10th Revision, Clinical Modification; CDC/BPA: Centers for Disease Control and Prevention/British Pediatric Association Classification of Diseases; TA: truncus arteriosus; TGA: transposition of the great arteries; d-TGA: d-TGA dextro-transposition of great arteries; TOF: tetralogy of Fallot; TAPVC: total anomalous pulmonary venous connection; IAA: interrupted aortic arch; DORV: double outlet right ventricle.

[a]Eye and ear anomalies are the focus of this year's data brief.

[b]These sub-categories of the broader reported conditions (transposition of the great arteries, pulmonary valve atresia and stenosis, and tricuspid valve atresia and stenosis) are presented given the interest in monitoring critical congenital heart defects targeted for pulse oximetry screening (Mai et al., 2012).

TABLE 2

Eye and ear anomalies counts, prevalence and 95% confidence interval for 30 U.S. population-based surveillance programs,[a] 2011–2015 (prevalence per 10,000 live births)

	Anophthalmia/microphthalmia[b]			Congenital cataract[c]			Anotia/microtia		
Variables	***n***	**prev**	**95% CI**	***n***	**prev**	**95% CI**	***n***	**prev**	**95% CI**
Total	1,785	1.5	1.4–1.5	1,818	1.5	1.4–1.6	2,2Q6	1.8	1.7–1.8
Case ascertainment methodology									
Active case-finding	951	1.9	1.8–2.1	776	1.6	1.5–1.8	1,282	2.6	2.5–2.7
Passive case-finding	834	1.2	1.1–1.2	1,042	1.4	1.3–1.5	924	1.2	1.2–1.3
Maternal race/ethnicity									
White, non-Hispanic	801	1.2	1.2–1.3	930	1.4	1.3–1.5	778	1.2	1.1–1.3
Black, non-Hispanic	290	1.5	1.3–1.6	333	1.7	1.5–1.9	170	0.8	0.7–1.0
Hispanic	550	1.9	1.8–2.1	413	1.6	1.4–1.7	1,037	3.6	3.4–3.9
Asian or Pacific Islander, non-Hispanic	76	1.3	1.0–1.6	78	1.3	1.0–1.6	145	2.4	2.0–2.8
American Indian or Alaska Native, non-Hispanic	17	1.9	1.1–3.0	9	1.0	0.5–1.9	23	2.6	1.6–3.8
Maternal age (years)									
<20	150	1.7	1.4–2.0	132	1.5	1.3–1.8	162	1.8	1.5–2.1
20–24	408	1.5	1.3–1.6	384	1.4	1.3–1.6	480	1.7	1.6–1.9
25–29	429	1.3	1.1–1.4	484	1.4	1.3–1.6	593	1.7	1.6–1.8
30–34	397	1.3	1.2–1.4	444	1.4	1.3–1.6	534	1.7	1.5–1.8
35–39	248	1.7	1.5–1.9	263	1.8	1.6–2.0	300	2.0	1.8–2.2
40+	131	3.7	3.1–4.4	90	2.5	2.0–3.1	117	3.2	2.7–3.9
Infant sex									
Male	841	1.4	1.3–1.4	938	1.5	1.4–1.6	1,205	1.9	1.8–2.0
Female	930	1.6	1.5–1.7	875	1.5	1.4–1.6	985	1.6	1.5–1.7

Prev: Prevalence; CI: Confidence Interval.

Other/unknown not shown.

[a]Contributing programs (total live births = 12,441,847) by case-finding status:

Active case-finding: Arizona, Delaware, Georgia (Metropolitan Atlanta), Louisiana, Massachusetts, Minnesota, North Carolina, Oklahoma, Puerto Rico, South Caro-lina, Texas, Utah (*n* = 12, total live births = 4,933,448).

Passive case-finding: Florida, Illinois, Kansas, Kentucky, Maine, Maryland, Michigan, Missouri, Nebraska, Nevada, New Jersey, New York, Ohio, Rhode Island, Tennessee, Vermont, Virginia, West Virginia (n = 18, total live births = 7,508,399).

[b] Excludes Maine (2011–2012), Maryland (2011–2012), Vermont.

[c] Excludes Maine (2011–2012), Maryland (2011–2012), Puerto Rico, Vermont.

TABLE 3

Eye and ear anomalies by laterality, U.S. population-based surveillance programs,[a] 13 programs, 2011–2015

	Anophthalmia/microphthalmia			Congenital cataract[b]			Anotia/microtia		
Variables	***n***	**(%)**	**95% CI**	***n***	**(%)**	**95% CI**	***n***	**(%)**	**95% CI**
Total[c]	982	100.0	-	648	100.0	-	1,316	100.0	-
Unilateral—total	380	38.7	35.6–41.8	319	49.2	45.3–53.2	1,039	79.0	76.6–81.1
Unilateral, left	178	18.1	15.8–20.7	147	22.7	19.5–26.1	388	29.5	27.0–32.0
Unilateral, right	192	19.6	17.1–22.2	171	26.4	23.0–30.0	648	49.2	46.5–52.0
Unilateral, not specified	10	1.0	0.5–1.9	1	0.2	0.0–0.9	3	0.2	0.0–0.7
Bilateral	602	61.3	58.2–64.4	329	50.8	46.8–54.7	277	21.0	18.9–23.4

CI: Confidence Interval.

[a]Participating programs: Arizona, Delaware, Georgia (Metropolitan Atlanta), Illinois, Louisiana, Massachusetts, North Carolina, Oklahoma, Puerto Rico, Rhode Island, South Carolina, Texas, Utah.

[b]Excludes Puerto Rico.

[c]Unknown/unspecified laterality is not shown: 11.1% of anophthalmia/microphthalmia cases, 23.0% of congenital cataract cases (includes anterior polar), 6.1% of anotia/microtia cases.

TABLE 4

Co-occurring birth defects by organ system for eye and ear anomalies from 30 U.S. population-based surveillance programs,[a] 2011–2015

	Anophthalmia/microphthalmia[b] (*n* = 1,798)			Congenital cataract[c] (*n* = 1,848)			Anotia/microtia (*n* = 2,217)		
Variables	***n***	**(%)**	**95% CI**	***n***	**(%)**	**95% CI**	***n***	**(%)**	**95% CI**
Congenital anomalies (740–759)/congenital malformations, deformations and chromosomal abnormalities (Q00-Q99)[d]	1,322	74.1	72.0–76.1	652	35.9	33.7–38.1	1,182	53.6	51.5–55.7
Central Nervous System (740–742, Q00–07)	657	36.8	34.6–39.1	194	10.7	9.3–12.2	251	11.4	10.1–12.8
Eye (743, Q10–15)[e]	-	-	-	-	-	-	286	13.0	11.6–14.4
Ear, Face, Neck (744, Q16–18)[e]	611	34.2	32.0–36.5	140	7.7	6.5–9.0	-	-	-
Cardiovascular (745–747, Q20–28)	767	43.0	40.7–45.3	342	18.8	17.0–20.7	611	27.7	25.8–29.6
Respiratory (748, Q30–34)	404	22.6	20.7–24.6	113	6.2	5.1–7.4	227	10.3	9.1–11.6
Orofacial Clefts (749, Q35–37)	305	17.1	15.4–18.9	42	2.3	1.7–3.1	186	8.4	7.3–9.7
Upper Gastrointestinal (750, Q38–40)	256	14.3	12.7–16.1	61	3.4	2.6–4.3	207	9.4	8.2–10.7
Lower Gastrointestinal (751, Q41–45)	141	7.9	6.7–9.2	47	2.6	1.9–3.4	110	5.0	4.1–6.0
Genital (752, Q50–56)	298	16.7	15.0–18.5	97	5.3	4.3–6.5	181	8.2	7.1–9.4
Renal (753, Q60–64)	293	16.4	14.7–18.2	103	5.7	4.6–6.8	306	13.9	12.5–15.4
Musculoskeletal (754, Q65–68)	449	25.2	23.2–27.2	111	6.1	5.0–7.3	313	14.2	12.8–15.7
Limbs (755, Q69–74)	516	28.9	26.8–31.1	118	6.5	5.4–7.7	260	11.8	10.5–13.2
Other Musculoskeletal (756, Q75–79)	525	29.4	27.3–31.6	136	7.5	6.3–8.8	513	23.3	21.5–25.1
Skin (757, Q80–84)	317	17.8	16.0–19.6	101	5.6	4.5–6.7	201	9.1	7.9–10.4
Chromosomal (758, Q90–99)	520	29.1	27.0–31.3	176	9.7	8.4–11.1	184	8.3	7.2–9.6
Other (759, Q85–89)	248	13.9	12.3–15.6	86	4.7	3.8–5.8	171	7.8	6.7–8.9

CI: Confidence Interval.

[a] Participating programs: Arizona, Delaware, Florida, Georgia (Metropolitan Atlanta), Illinois, Kansas, Kentucky, Louisiana, Maine, Maryland, Massachusetts, Michigan, Minnesota, Missouri, Nebraska, Nevada, New Jersey, New York, North Carolina, Ohio, Oklahoma, Puerto Rico, Rhode Island, South Carolina, Tennessee, Texas, Utah, Vermont, Virginia, West Virginia (total live births = 12,441,847).

[b] Excludes Maine (2011–2012), Maryland (2011–2012), Vermont.

[c] Excludes Maine (2011–2012), Maryland (2011–2012), Puerto Rico, Vermont.

[d] Birth defects that fall outside the 740–759 range for ICD-9-CM and/or CDC/BPA or outside the Q00-Q99 range for ICD-10-CM were not examined. Cases are counted separately for each organ system (categories are not mutually exclusive).

[e] Co-occurring birth defects within the same organ system for the conditions of interest (eye and ear anomalies) are not presented given our inability to ensure diagnosis codes are unrelated to the conditions of interest.