Making Healthy Choices to Prevent Birth Defects - Make a PACT for Prevention

National Birth Defects Prevention Month occurs each January. The National Birth Defects Prevention Network’s (NBDPN) goal for promoting this month-long event is to build upon the 2014 theme that birth defects are “Common, Costly, and Critical,” and to offer practicable, preventive steps that can be taken by professionals, community groups, and the public. While not all birth defects can be prevented, steps can be taken to increase a woman’s chance of having a healthy baby. This year’s theme for National Birth Defects Prevention Month is, “Making Healthy Choices to Prevent Birth Defects - Make a PACT for Prevention.”

The NBDPN provides free resources and materials promoting birth defects awareness and prevention. Previous years’ themes have included “Birth Defects are Common, Costly, and Critical” (2013 and 2014), “And the Beat Goes On...Looking to the Future for Healthy Hearts” (2012), and “Medication Use Before, During, and After Pregnancy” (2011).

For more information, and to access these resources, visit http://www.nbdpn.org/national_birth_defects_prevent.php. This January, let’s make a PACT for prevention!

PACT:
♦ Plan Ahead
♦ Avoid harmful substances
♦ Choose a healthy lifestyle
♦ Talk to your doctor

World Birth Defects Day will be celebrated on March 3, 2015. This will be the first time that a global day promoting birth defects awareness and prevention will be observed. Organizations in the U.S. involved with the promotion of this day include the NBDPN, CDC, March of Dimes, and International Clearinghouse for Birth Defects Surveillance and Research.
Updated Texas Birth Defects Registry Data, 1999-2011

The Texas Birth Defects Registry has released its Report of Birth Defects Among 1999-2011 Deliveries. The full report shows the birth prevalence of selected defects among deliveries to Texas residents during 1999-2011 by year, mother’s age, mother’s race/ethnicity, sex of the infant or fetus, and region of the state, as well as the pregnancy outcome distribution for selected birth defects. Some of the time trends and maternal age patterns included in the full report are highlighted below.

Between 1999 and 2011, about 428 babies with a structural or chromosomal birth defect were born per every 10,000 live births (or 4.3%) to Texas residents. The most common birth defect was atrial septal defect, a heart defect with a prevalence of 59 cases per 10,000 live births (0.6%, or one in every 169 births). In fact, five of the 15 leading birth defects involve the cardiovascular system, as shown in the figure below.

![The 15 Most Common Birth Defects in Texas, 1999-2011](image)

**Prevalence of Birth Defects Over Time, 1999-2011**

Overall, 3.3% (about 332 cases per 10,000 live births) of all babies in 1999 were born with one or more structural malformations or chromosomal anomalies. By 2011, this rose to 5.5% (about 553 cases per 10,000 live births). Although the overall prevalence of birth defects appears to be increasing with time, not all birth defects follow this trend. The chart at the top of the next page shows time trends from 1999 to 2011 for three different birth defects: gastroschisis, spina bifida, and hypoplastic left heart syndrome.

Langlois et al. (2011) recently studied time trends for birth defects in Texas from 1999-2007. In this analysis, the authors found a 3.6% increase in the prevalence of total birth defects per year. Increases remained significant after adjustment for demographic characteristics. The increases in prevalence were the largest in regions where variations in diagnostic practices were more common, showing that the overall increases may not be due to increased occurrence, but rather due to increased awareness, documentation in health care facilities, and detection of certain birth defects in the first year of life. Gastroschisis is an example of an important birth defect with increasing prevalence over time.

A basic question regarding birth defects is whether there is an association between the prevalence of birth defects and mother’s age. Data from the Texas Birth Defects Registry provides insight on this matter. In Texas from 1999-2011, for any monitored birth defect, the prevalence of birth defects in babies born to mothers over the age of 30 increased with each increasing five-year maternal age group. An example of a specific birth defect that seems to occur more in babies born to older mothers is Trisomy 21, or Down syndrome. For babies born to mothers age 40+, the prevalence of Down syndrome was about 122/10,000 live births; while the prevalence was about 7/10,000 among births to mothers under 20 years of age.

While many defects occurred more among births to older mothers, there were some defects, such as pyloric stenosis, that were more common among births to younger mothers. Women less than 20 years of age were about twice as likely to have a baby with pyloric stenosis compared to women age 40+. Another notable difference was in the prevalence of gastroschisis among births in women <20 compared to births in older women. There were about 16 babies per 10,000 live births born among young mothers with gastroschisis and only about one per 10,000 live births among older mothers. The figures below show the prevalence of Down syndrome and gastroschisis by maternal age in Texas from 1999-2011.

Maternal Age and Prevalence of Birth Defects, 1999-2011

To view the complete Annual Report, visit http://www.dshs.state.tx.us/birthdefects/BD_data.shtm
Survival Patterns of Selected Congenital Heart Defects

Congenital heart defects are conditions present at birth that affect the structure and function of the heart. They are one of the most common types of birth defects, affecting approximately 1% of all births. Texas researchers and collaborators have conducted several registry-based studies looking at the mortality and survival patterns for major heart defects, including critical congenital heart defects (CCHDs). Key findings are found in the box below, followed by more detailed summaries from each contributing article. Hazard Ratios (HRs) indicate risk of death, adjusting for important cofactors.

<table>
<thead>
<tr>
<th>Key Findings</th>
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<tbody>
<tr>
<td>• Non-Hispanic black children in Texas had a higher risk of death from several important heart defects, compared to non-Hispanic white children</td>
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<tr>
<td>• Texas children had a higher risk of death from specific heart defects if they:</td>
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<tr>
<td>• were born in an earlier time period (1996-2000 vs. 2001-2003)</td>
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<tr>
<td>• had co-occurring extracardiac defects</td>
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<tr>
<td>• Among the major heart defects, hypoplastic left heart syndrome had the lowest probability of survival in Texas children (roughly 1/3 survived five years)</td>
</tr>
<tr>
<td>• For Texas babies born with hypoplastic left heart syndrome:</td>
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<tr>
<td>• pre-surgical mortality was highest for those with greatest driving distance from the birth hospital to a cardiac surgical center</td>
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<tr>
<td>• post-surgical mortality was lower for cardiac specialty centers with higher patient volume</td>
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</table>

Mortality in First Five Years in Infants with Functional Single Ventricle Born in Texas, 1996-2003


This study looked at five-year survival and factors associated with survival for infants born with functional single ventricle. Infants with hypoplastic left heart syndrome, single ventricle complex, pulmonary atresia with intact ventricular septum, and tricuspid atresia born between 1996 and 2003 were included in the analysis. The authors concluded that some racial/ethnic groups had a higher risk of death than others, and that defect type, birth era, and having extracardiac defects also affected the risk of death.

• Five-year survival (%) was different for each defect type:
  • Hypoplastic left heart syndrome, 38.0%
  • Single ventricle, 56.1%
  • Pulmonary atresia intact ventricular septum, 55.7%
  • Tricuspid atresia, 74.6%

• Race/Ethnicity:
  • Non-Hispanic blacks had a 41% higher risk of death than NH whites (adj. HR=1.41, 95% CI=1.01-1.97)
  • Hispanics had a 26% higher risk of death (adj. HR=1.26, 95% CI=1.00-1.58)

• Birth Era: 47% lower risk of death for patients born 2001-03 vs. 1996-2000 (adj. HR=0.53, 95% CI=0.43-0.66)

• Extracardiac Defects: 84% higher risk of death with extracardiac defects (adj. HR=1.84, 95% CI=1.46-2.34)
Racial/Ethnic Disparities in Risk of Early Childhood Mortality Among Children with Congenital Heart Defects


For this study, researchers examined the relationship between maternal race/ethnicity and early childhood mortality in Texas children who were born with selected congenital heart defects. The study included live-born infants with a CHD, born between 1996 and 2003, and looked at deaths occurring between 1996 and 2005. They found that for certain conditions, some racial/ethnic groups had a higher risk of early childhood mortality.

✦ Higher risk of death in non-Hispanic blacks (compared to NH whites) for:
  - Transposition of the great arteries (adj. HR*=2.04, 95% CI=1.40-2.97)
  - Tetralogy of Fallot (adj. HR=1.85, 95% CI=1.09-3.12)
  - Pulmonary valve atresia without ventricular septal defect (adj. HR=2.60, 95% CI=1.32-5.12)
  - Ventricular septal defect (adj. HR=1.56, 95% CI=1.19-2.03)
  - Atrial septal defect (adj. HR=1.34, 95% CI=1.08-1.66)

✦ Higher risk of death in Hispanics (compared to NH whites) for:
  - Pulmonary valve atresia w/o VSD (adj. HR=1.76, 95% CI=1.06-2.91)
  - Hypoplastic left heart syndrome (adj. HR=1.51, 95% CI=1.13-2.02)

(*Hazard Ratios (HRs) were adjusted for maternal age, education, infant gender, border county residence, rural urban commuting area, number of defects, birth weight/gestational age combinations.)

Effect of Acculturation and Distance from Cardiac Center on Congenital Heart Disease Mortality


This study looked at the association between acculturation (specifically, having a Latin American-born parent) and first-year mortality in infants born between 1996 and 2003 with CCHD. The authors also examined the association between distance from maternal residence to a cardiac center and first-year mortality in infants with CCHD. The authors found:

✦ For eight severe heart defects combined (hypoplastic left heart syndrome (HLHS), Ebstein’s malformation of the tricuspid valve, interrupted aortic arch, truncus arteriosus, pulmonary atresia and intact ventricular septum, pulmonary valve atresia with ventricular septal defect, tricuspid atresia, and single ventricle):
  - First year survival was 59.9%.
  - There were no racial/ethnic differences in survival.
  - There was lower survival among residents of Texas counties bordering Mexico.
  - Neither parental birth country (Latin American vs. US-born) nor distance from home to cardiac center was significantly related to first-year survival.
  - Infant birth weight, gestational age, and extracardiac defects were risk factors for mortality.

✦ Lower survival in Hispanics (compared to NH whites, p<0.05) for HLHS

(Continued on page 6)

February 14 is Congenital Heart Defect Awareness Day. Congenital heart defects are the most common type of birth defect, affecting about 40,000 babies per year in the U.S.
Age at Referral and Mortality from Critical Congenital Heart Disease

Fixer DE, Xu P, Nembhard WN, Ethen MK, and Canfield MA. Age at Referral and Mortality From Critical Congenital Heart Disease. Pediatrics, 2014; 134(1), e8-e105.

This study evaluated the association between time to referral to a cardiac center and neonatal mortality in 2,360 Texas neonates with Critical Congenital Heart Disease (CCHD) born between 1996 and 2007, before pulse oximetry screening was implemented in Texas.

- **Increased postneonatal mortality was found for non-Hispanic blacks (as compared to non-Hispanic whites) for transposition of the great arteries (HR=2.4; 95% CI, 1.5-4.0), pulmonary valve atresia without ventricular septal defect (HR=4.1; 95% CI 1.7-9.7), Ebstein’s anomaly (HR=8.6; 95% CI, 1.2-61.1), hypoplastic left heart syndrome (HR=2.1, 95% CI, 1.2-3.7), coarctation of the aorta (HR=2.1; 95% CI 1.2-3.5), ventricular septal defect (HR=2.1; 95% CI, 1.6-2.8), and atrial septal defect (HR=1.4; 95% CI, 1.1-1.8).**

- **Increased postneonatal mortality risk was also found for tetralogy of Fallot in Hispanics, as compared to non-Hispanic whites (HR=2.0; 95% CI, 1.1-3.5).**

- **An increase in mortality risk during early childhood (after one year of age) was observed for non-Hispanic blacks for tetralogy of Fallot (HR=3.1; 95% CI, 1.1-9.1), tricuspid valve atresia/stenosis (HR = 12.8; 95% CI 1.7-95.5), and ventricular septal defect (HR=2.4; 95% CI, 1.4-4.1).**

Prenatal Diagnosis, Birth Location, Surgical Center, and Neonatal Mortality in Infants with Hypoplastic Left Heart Syndrome


In this recent study, the first of its kind, researchers evaluated the effect of calculated driving time from birth center to a cardiac surgical center (CSC) on neonatal mortality among infants with hypoplastic left heart syndrome (HLH), and evaluated the associations between prenatal diagnosis, CSC HLHS
volume, and mortality. This retrospective, population-based study used nine years of data (1999-2007) from the Texas Birth Defects Registry. This study found:

- A significant increase in neonatal mortality with increasing driving time from birth center to a cardiac surgical center (CSC). For infants born less than 10 minutes from a CSC, neonatal mortality (death before 28 days of age) was 21.0%. For those born between 10 and 90 minutes from a CSC, neonatal mortality was 25.2%, and for those born more than 90 minutes away, neonatal mortality was 39.6% (p value for trend <0.001).
- Infants who were born more than 90 minutes from a CSC had two-fold higher odds of neonatal mortality as compared to infants born less than 10 minutes from a CSC (adjusted odds ratio (OR), 2.03; 95% confidence interval (CI), 1.19-3.45).
- There was no significant association between prenatal diagnosis alone and improved survival among infants (p=0.14).
- Infants born more than 90 minutes from a CSC had a much higher pre-transport mortality (adjusted OR, 6.69; 95% CI, 2.52-17.74).
- Lower surgical mortality was associated with higher CSC HLHS volume (adjusted OR of 0.88 per 10 HLHS patients; 95% CI, 0.84-0.91). Therefore, for every 10 additional HLHS patients seen in a CSC, the odds of neonatal death decreased by about 12%.

This study also suggests that planning of delivery near a large volume Cardiac Surgical Center may improve survival among infants with HLHS. In order to plan delivery near a large hospital, improving prenatal diagnosis is important.

Recent Publications


(Continued on page 8)
Quality Assurance Activities of the Texas Birth Defects Registry

The Texas Birth Defects Registry, a population-based, active surveillance system, utilizes multiple quality assurance activities to ensure a high quality of work by its data collection staff, including re-casefinding, re-review of medical records, re-abstraction, data entry review, field review, clinical review, diagnosis code review, and evaluation of timeliness. Data generated by these activities are used for multiple purposes such as evaluating data collection over time and across geographic regions to identify possible inconsistencies, evaluating job performance of surveillance specialists, and identifying reoccurring problem areas where staff might need more training, better procedures, or where enhancements to the system may be needed.

In general, the error rates measured by most of these quality assurance activities and the timeliness of the data have improved over time. One of the most important measures is the timeliness measure, which allows a comparison of a program’s timeliness with the national benchmark established by the National Birth Defects Prevention Network (NBDPN). The Texas Birth Defects Registry has observed a significant overall improvement in timeliness from delivery year 2003 to 2010. Another important measure is diagnosis code review, which provides an unbiased look at the quality of records entered into the system, as well as an evaluation of consistency between clinical reviewers. A random selection of 10% of records assigned for diagnosis code review revealed significant improvement over time, with a decline in the percentage of completed abstraction records returned for corrections from 20% in 2003 to 7% in both 2011 and 2012.
**Texas Scores Well on National Data Quality Assessment**

The National Birth Defects Prevention Network’s (NBDPN) data quality assessment evaluates the completeness, timeliness, and accuracy of data for population-based birth defects surveillance programs. They evaluate the programs based on eleven self-reported indicators of data quality. Three levels of performance are associated with each data quality measure: level one signifies a rudimentary level of performance, level two signifies an essential level of performance, and level three represents an optimal level of performance by a surveillance program. An essential level of performance is expected to be met by each program for all indicators. For the 2014 transition year, Texas met or exceeded all expectations for each indicator of data quality, and achieved an optimal level of performance for each data quality indicator of accuracy. In addition, Texas scored above the national average for each category (completeness, timeliness, and accuracy). The results of Texas’ performance measure scores are shown below.

![Performance Measure Scores, Texas, 2014](image)

**Data Quality Measures**

- **DQ1.1** Types of data sources used systematically and routinely to identify potential cases at a population-based level
- **DQ1.2** Birth defects included using standard case definitions
- **DQ1.3** Pregnancy outcomes included
- **DQ1.4** Systematic & routine identification of cases during an ascertainment period (age at diagnosis)
- **DQ1.5** Data elements collected
- **DQ2.1.1** Time of case data completion for NBDPN core list
- **DQ2.1.2** Time of case data completion for NBDPN recommended list
- **DQ3.1** Data quality procedures for verification of cases diagnosis
- **DQ3.2** Scope of birth defect verified
- **DQ3.3** Level of expertise for individuals who perform case diagnosis verification
- **DQ3.4** Database quality assurance process
Recent publications continued


Treating for Two: Safer Medication Use in Pregnancy

Treating for Two: Safer Medication Use in Pregnancy, is a new CDC initiative aiming to improve maternal health and prevent birth defects by identifying the best treatments for common conditions during childbearing years, during pregnancy, and while breastfeeding.

Before pregnancy: If you are thinking about becoming pregnant, talk with your doctor about medications you are currently taking and about other steps you can take in preventing birth defects.

During pregnancy: Talk to your doctor about medications and supplements you are taking or thinking about taking. Don’t stop or start any medications before talking with your doctor.

Visit the Treating for Two website:

www.cdc.gov/pregnancy/meds/treatingfortwo/

Key Findings: Racial and Ethnic Differences in the Occurrence of Major Birth Defects

The American Journal of Public Health published a new study that examined the occurrence of major birth defects across multiple racial and ethnic groups. Using 12 state-based birth defects tracking systems, this is to date the largest study conducted to look at racial and ethnic differences in the United States for a range of birth defects.

Main Findings from this Study

♦ Compared with non-Hispanic whites, American Indians/Alaska Natives had a significantly higher occurrence of:
  • Anotia/Microtia (ear defects)
  • Cleft lip with or without cleft palate
  • Trisomy 18
  • Encephalocele (serious defect of the skull and brain)
  • Limb deficiency (when part or all of the arm or leg fails to form completely during pregnancy)

♦ Cubans and Asians, especially Chinese and Asian Indians, had significantly lower occurrence of many of the studied birth defects, compared to non-Hispanic whites.

♦ Some of the studied birth defects had very different rates of occurrence across the racial and ethnic groups (either much higher or much lower occurrence compared to non-Hispanic whites). Conditions that showed substantial variation across the racial and ethnic groups include anotia/microtia, spina bifida (a defect of the spine), and Down syndrome.

♦ The researchers recommend further investigation to determine why there appeared to be a relatively high occurrence of certain birth defects in American Indians/Alaska Natives.
Birth defects that have significantly higher or lower occurrence in selected racial and ethnic groups, compared to non-Hispanic white

<table>
<thead>
<tr>
<th>Mother’s Race/Ethnicity</th>
<th>Much Higher Occurrence1</th>
<th>Much Lower Occurrence2</th>
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<tbody>
<tr>
<td>Non-Hispanic American Indian/Alaskan Native</td>
<td>Encephalocele</td>
<td>Hypospadias</td>
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<td></td>
<td>Anotia/Microtia</td>
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<td></td>
<td>Cleft lip with or without cleft palate</td>
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<td></td>
<td>Upper limb deficiency</td>
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<td></td>
<td>Any limb deficiency</td>
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<td></td>
<td>Trisomy 18</td>
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<tr>
<td>Non-Hispanic Asian</td>
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<td>Spina bifida without anencephaly</td>
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<td>Truncus arteriosus</td>
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<td>Aortic valve stenosis</td>
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<td>Hypoplastic left heart syndrome</td>
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<td>Coarctation of the aorta</td>
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<td>Pyloric stenosis</td>
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<td>Hypospadias</td>
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<td>Gastrochisis</td>
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<td>Omphalocele</td>
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<tr>
<td>Non-Hispanic Black</td>
<td>Encephalocele</td>
<td>Aortic valve stenosis</td>
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<tr>
<td></td>
<td>Trisomy 18</td>
<td>Cleft lip with or without cleft palate</td>
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<td>Pyloric stenosis</td>
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<td>Gastrochisis</td>
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<td>Hispanic</td>
<td>Anencephaly</td>
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<td>Encephalocele</td>
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<td></td>
<td>Anotia/Microtia</td>
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</table>

1 These conditions were found to have statistically significantly higher occurrences in the noted races and ethnicities compared to non-Hispanic white
2 These conditions were found to have statistically significantly lower occurrences in the noted races and ethnicities compared to non-Hispanic white.
Note: Even though programs try to collect information on all occurrences, some birth defects might not be captured by programs if the outcome is not a live birth. This could underestimate the occurrences of some birth defects.

About this Study

- Researchers used data from 12 population-based birth defects tracking programs. Population-based means that the researchers look at all babies with birth defects who live in a defined study area, in order to get a complete picture of what is happening among all of the people (the population) living in the study area. Participating states were: Arizona, Colorado, Florida, Georgia (some counties), Illinois, Massachusetts, New York, North Carolina, and Texas. This study included 13.5 million live U.S. births from 1999 to 2007, meaning 1 of every 3 births in the United States during that time period was included in the study.

- This is the most comprehensive study to date to examine racial and ethnic variation for a wide range of birth defects in the United States, particularly among under-studied racial and ethnic groups including American Indians/Alaska natives, Hispanic subgroups, and Asian subgroups.


The Monitor is published annually by the Birth Defects Epidemiology and Surveillance Branch, Texas Department of State Health Services.

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Please visit the BDES website for updated information: www.dshs.state.tx.us/birthdefects.

Request for additional copies or back issues may be made to birthdefects@dshs.state.tx.us.

2015 Calendar

- January: National Birth Defects Prevention Month
- January 5-11: National Folic Acid Awareness Week
- January 24-27: Association of Maternal and Child Health Programs (AMCHP) Annual Conference, Washington, DC
- February: American Heart Month
- February 2-7: Society for Maternal-Fetal Medicine Annual Meeting
- February 6-9: 15th Annual International Symposium on Congenital Heart Disease, St. Petersburg, Florida
- February 14: Congenital Heart Defect Awareness Day
- Spring 2014: March of Dimes March for Babies (check with MOD for specific dates and locations)
- March 3: World Birth Defects Day
- March 4-7: 6th International Conference on Fetal Alcohol Spectrum Disorder, Vancouver, BC, Canada
- April: National Autism Awareness Month
- April 6-12: National Public Health Week, American Public Health
- May 3: Parents of Preemies Day
- June 27-July 1: 54th Annual Meeting of the Teratology Society, Montreal, QC, Canada
- July: National Cleft and Craniofacial Awareness & Prevention Month
- July: Gastroschisis Awareness Month
- September: Childhood Cancer Awareness Month
- September: Newborn Screening Awareness Month
- October: National Down Syndrome Awareness Month

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