

Birth defects are health conditions present at birth that can change how the body works. They are the leading cause of infant death. While many factors cause birth defects, steps can be taken to help prevent or limit certain risks, such as exposure to air pollution and chemicals, use of alcohol and drugs, a lack of folic acid in the diet and no prenatal care. The Centers for Disease Control and Prevention reports that 1 in 33 U.S. babies has a birth defect, which is about 4,200 babies born with birth defects in Pennsylvania each year.

HOW TO LIMIT THE RISK OF BIRTH DEFECTS BEFORE/DURING PREGNANCY

- Take a prenatal multivitamin with folic acid every day along with eating a healthy diet.
- Do not smoke and avoid alcohol, drugs, and toxic exposures.
- Prevent infections during pregnancy, e.g., Zika.
- Work with a health care provider to control chronic medical conditions, e.g., diabetes.
- Consult a health care provider to learn which medications are safe for pregnancy.
- Visit a doctor regularly for prenatal care and to discuss family health history and genetic testing.



MONITORING BIRTH DEFECTS IN PENNSYLVANIA

Pennsylvania does not have a statewide birth defects registry. However, the state tracks 10 birth defects (below). Data are on the [Enterprise Data Dissemination Informatics Exchange¹](#) by county and come from the Pennsylvania Vital Statistics. They reflect conditions diagnosed at birth and noted on the birth record.

- Anencephaly is a fatal neural tube defect present at birth. The baby is born without parts of the brain or skull bones.
- Cleft lip and Cleft palate are orofacial clefts that occur when the upper lip or palate or both do not form completely.
- Congenital diaphragmatic hernia (CDH) occurs when the diaphragm muscle fails to close and organs from the abdomen move into the chest.
- Congenital heart defects affect the structure of a baby's heart and the way it works. They are the most common birth defect. Pennsylvania tracks cyanotic congenital heart disease, specifically.
- Down Syndrome/Trisomy 21 is a chromosomal disorder that includes a combination of birth defects. Chromosomes are structures in cells that contain genes.
- Gastroschisis occurs when an infant's abdominal organs stick out of the body through a hole near one side of the belly button and do not have a protective sac.
- Hypospadias occurs when a male baby's urethral opening is located at the underside and not the tip of the penis.
- Limb reduction defects occur when part of or the entire limb of a baby fails to grow completely during pregnancy.
- Omphalocele occurs when an infant's abdominal organs stick out of the body through a hole in the belly button. Unlike gastroschisis, the organs do have a protective sac.
- Spina bifida refers to a group of neural tube defects that affects the spine and spinal cord. Pennsylvania tracks meningocele, the most severe form.

BIRTH DEFECTS PROGRAMS IN PENNSYLVANIA

The [Newborn Screening Program²](#) screens newborns in Pennsylvania for hearing loss, congenital heart defects, metabolic and other genetic conditions. As the baby grows, the [Special Kids Network³](#) (SKN) may be a useful resource for families, caregivers and health care providers who work with children with special needs.

If you have questions or concerns about birth defects, please contact the Division of Environmental Health Epidemiology at dehe@pa.gov or 717-787-3350.

August 2023

¹ <https://www.health.pa.gov/topics/HealthStatistics/EDDIE/Pages/EDDIE.aspx>

² <https://www.health.pa.gov/topics/programs/Newborn-Screening/Pages/Newborn%20Screening.aspx>

³ <https://www.health.pa.gov/topics/programs/Special%20Kids%20Network/Pages/Special%20Kids%20Network.aspx>