According to the Centers for Disease Control and Prevention (CDC), congenital anomalies, or birth defects, are common and affect about one in every 33 babies born in the United States each year. Furthermore, birth defects are the leading cause of infant deaths and account for roughly 20 percent of all infant deaths in the United States. Birth defects can affect almost any part of the body, range from mild to severe, and affect how the body looks, works or both. Some birth defects are harmless, while others require long-term medical treatment. Birth defects can occur during any stage of pregnancy. Most occur in the first three months of pregnancy when the organs of the baby are forming; however, some defects occur later in pregnancy while the tissues and organs are continuing to grow and develop.

During pregnancy, screening and diagnostic tests (which include blood work, ultrasounds and testing of the amniotic fluid) should be performed and can identify birth defects like neural tube defects or chromosomal disorders, such as Down syndrome (Trisomy 21). Certain birth defects, such as cleft lip or cleft palate, are usually diagnosed at the time of birth (although they can be seen on ultrasounds before birth), while other birth defects, such as heart defects and hearing loss, may not be diagnosed until later after a baby is born.

This article highlights the data collected for Pennsylvania resident live births, which are presented in our publications and on the EDDIE data tool. Not all types of congenital anomalies are collected for resident births, only those as listed on the Pennsylvania certificate of live birth (2003 revised version). Many types of birth defects are discovered later, after birth. Therefore, these data do not represent all types of congenital anomalies that occur. The birth defects included here are identified in less than 1 percent of all births in Pennsylvania during a given year. As shown on Table 1, out of the 140,727 live births in 2015 for Pennsylvania resident live births, 620 births had one of the listed congenital anomalies.
residents, 620 births reported having a congenital anomaly reportable through the certificate of birth. While this may be a small percentage of overall births, the impact on newborns and their families can be very significant.

Many complex factors can contribute to birth defects. These include genetics, behaviors and pre-existing medical conditions, and environmental influences or exposures. We know the cause for some birth defects, but, for most, we do not. Per the CDC, certain factors might increase the chances of having a baby with a birth defect, such as:

- women who smoke, drink alcohol or take certain drugs;
- women with certain medical conditions, such as diabetes or obesity;
- taking certain medications known to cause birth defects, such as isotretinoin, which is used to treat severe acne;
- being an older mother, typically 35 years of age and older; and
- having a family history of birth defects.

To learn more about your risk of having a baby with a birth defect, you can talk with a clinical geneticist or a genetic counselor. Health care providers can help their patients find a counselor or geneticist in their area, and a medical school or university medical center may have information about finding a genetic professional.

Serious medical conditions a mother experiences before or during pregnancy, as well as complications during labor and delivery, affected some of the 620 births resulting in birth defects in Pennsylvania (see Chart 1). The most common medical conditions reported were risk factors present during the pregnancy. Risk factors in pregnancy reported on the certificate of birth include diabetes, hypertension, vaginal bleeding during pregnancy before the onset of labor and pregnancy resulting from infertility treatment. Previous pre-term births, cesarean sections or poor pregnancy outcomes are also risk factors. Of the 620 births resulting in one or more of the selected congenital anomalies, 223 women experienced risk factors during pregnancy, and about 70 percent were less than 35 years old, while 30 percent were 35 and older.

Infections during pregnancy and maternal morbidity are two medical conditions that were rarely reported on birth certificates for births in Pennsylvania that resulted in a congenital anomaly. Infections recorded on birth certificate reporting include gonorrhea, syphilis, herpes simplex virus, chlamydia, hepatitis B, and hepatitis C. Examples of maternal morbidity include maternal blood transfusions, third or fourth degree perineal lacerations, tearing of the uterine wall, unplanned hysterectomy, admission to an intensive care unit, or an unplanned operating room procedure following delivery.

About 60 percent of children born in Pennsylvania with one of these selected birth defects experience an abnormal condition of the newborn as reported on the birth
For 378 infants out of the 620 with a recorded birth defect, additional medical support, such as assisted ventilation (excluding oxygen only and laryngoscopy for aspiration of meconium) or additional care in the NICU (neonatal intensive care unit), may have been required. Some infants need antibiotics or surfactant replacement therapy. Others suffer from seizures or serious neurologic dysfunction.

Lifestyle choices, such as smoking, also negatively affect pregnancy and are known contributing factors to birth defects. In Pennsylvania, about one out of every six mothers that gave birth to a child with a birth defect smoked during pregnancy, with most of those being younger mothers (see Chart 2, previous page). Mothers younger than 35 years old were more likely to smoke than mothers ages 35 and older. The data shows that Pennsylvania resident mothers under 25 years of age were 1.7 times more likely to smoke than mothers 35 years of age and older. And resident mothers between 25 and 34 years old were well over two times (2.3) as likely to smoke than mothers 35 and older.

Many birth defects cannot be prevented, but there are some ways to lower the risk of having a baby with a birth defect. Women should visit their doctor regularly and start prenatal care if they are planning for a future pregnancy or as soon as they think they may be pregnant. Recommendations include taking 400 mcg (micrograms) of folic acid daily and avoiding other medications (such as over-the-counter and supplements) without consulting a doctor first. In addition, avoiding alcohol, drugs and tobacco, maintaining a healthy weight, and controlling serious medical conditions (such as diabetes) may help reduce the risk of complications during pregnancy.

Babies with birth defects often need special care and interventions to survive and thrive developmentally. According to the CDC, state birth defects tracking programs provide one way to identify and refer children as early as possible for services they need. Early intervention is vital to improving outcomes for these babies. An infant’s doctor should be able to provide local resources and treatment options to best help affected children. Clinical geneticists, genetic counselors and other specialists are another great resource.

For additional information on birth defects, visit the Pennsylvania Department of Health’s Birth Defects page. Birth defect data among Pennsylvania residents is also available on EDDIE, the department’s data dissemination tool. Additional information can also be found on the Centers for Disease Control and Prevention Birth Defects site. For questions regarding the data and statistics presented in this article, please contact the Division of Health Informatics.