Kentucky Department for Public Health

Birth Defects Surveillance in Kentucky: 2015-2024

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Section 1: Executive Summary

About 1 in every 33 births in the United States (U.S.) is affected by a major birth defect that requires medical intervention or has serious impacts on health or development (CDC, 2025; NBDPN, 2004). In the U.S., birth defects are the leading cause of infant mortality, causing about 20% of all deaths before the first birthday (CDC, 2025). The purpose of this report is to establish Kentucky-specific data for birth defects, for the timeline of 2015-2024.

The Kentucky Birth Surveillance Registry is a state-mandated surveillance system that utilizes a web-based database to collect, link, and track information on children with birth defects from birth to age five. Cases are reported from the Kentucky Hospital Association (KHA), live birth certificate, death certificate, stillbirth certificate, Critical Congenital Heart Defect (CCHD) surveillance, outpatient clinics, and genetics laboratories.

There were 72,114 children with any birth defect in this 10-year period, of whom 20,235 had major birth defects. The rate of major birth defects is 380 per 10,000 live births, or approximately 1 in 26 live births, which is about 25% higher than national estimates (CDC, 2025). Overall, cardiovascular defects are the most common type of birth defect. The appendix of this report includes rates of all major birth defects, individually, with comparisons to previous Kentucky estimates and U.S. estimates whenever available.

In this sample, pre-existing maternal diabetes is the risk factor with the greatest association with any birth defects (four-fold increase in likelihood of neural tube defects and heart defects). However, maternal pre-pregnancy obesity (occurring in 32.9% of Kentucky mothers) and smoking during pregnancy (reported by 12.4% of Kentucky mothers) are much more common risk factors that are associated with increased occurrence of birth defects. Maternal age and hypertension are also explored in this report.

Furthermore, the connection between birth defects and preterm birth is described. Birth defects mortality is another concern, and this report includes information about manners and causes of death for children with birth defects, the most common types of birth defects to cause death or stillbirth (central nervous system anomalies and chromosomal anomalies), and birth defects with the highest fatality rates (anencephaly, Trisomy 13, and Trisomy 18).

In conclusion, this report emphasizes that birth defects are a common and critical concern in Kentucky, with a substantial impact on childhood morbidity and mortality. Supporting healthy pregnancies is one way to promote positive pregnancy outcomes, and this report is designed to inform public health practice and prevention initiatives.

Section 2: Background

What are birth defects?

The Centers for Disease Control and Prevention (CDC) define birth defects, or congenital anomalies, as health conditions that alter the structure of one or more body parts and occur during development in pregnancy (2025). Birth defects are typically classified as structural or functional/developmental. Structural defects involve physical changes to body parts, such as congenital heart defects or cleft lip and palate. Functional or developmental defects affect how the body works, such as chromosomal disorders. Some birth defects are minor and may require no interventions, while others may require surgery or supporting therapies. In severe cases, birth defects can result in lifelong disability or death (CDC, 2025). As birth defects vary so widely, and as the term "birth defect" and "congenital anomaly" can have negative implications, it is best to speak about specific conditions whenever possible (CDC, 2025).

About 1 in every 33 births in the United States (U.S.) is affected by a major birth defect that requires medical intervention or has serious impacts on health or development (CDC, 2025; NBDPN, 2004). In the U.S., birth defects are the leading cause of infant mortality, causing about 20% of all deaths before the first birthday (CDC, 2025). The following sections of this report will explore the prevalence of birth defects in Kentucky and how that differs from national metrics.

Prevention of birth defects

The causes of most birth defects are unknown and may be related to a complex mix of factors, including genetics, behaviors, and environmental exposures (CDC, 2025). Having any known risk factor does not mean a baby will be born with a birth defect, and babies can be born with a birth defect even in the absence of any known risk factors (CDC, 2025). However, there are ways to support healthy pregnancies and increase the chance of having a healthy baby. Recommendations include abstaining from: tobacco use, including cigarette smoking and use of electronic nicotine delivery systems (ENDS); alcohol use; and illicit drug use (CDC, 2025). Seeking health care provides the opportunity for guidance on managing chronic health conditions such as diabetes or high blood pressure; infections and acute illnesses causing fever; and medication use, including prescription, over-the-counter, and supplements (CDC, 2025). As part of prenatal care, clinicians can provide advice on nutrition, including folic acid and other vitamin needs, exercise, weight management, vaccinations, and more.

Most birth defects occur during the first three months of pregnancy, but they can occur at any stage (CDC, 2025). Identification of a birth defect could occur during pregnancy, at birth, in early childhood, or later in life (CDC, 2025). Some birth defects are visible upon examination, but others must be diagnosed with specialized tests or equipment (CDC, 2025). Accurate identification of birth defects is critical for planning to meet the child's medical and

developmental needs, and early engagement in appropriate care can lead to improved quality of life and reduced healthcare costs (CDC, 2025).

Kentucky Birth Surveillance Registry

The Kentucky Birth Surveillance Registry (KBSR) is a state-mandated surveillance system that started in 1998 to provide information on the incidence, prevalence, trends, and possible causes of stillbirths, birth defects, and disabling conditions. KBSR was established by Kentucky Revised Statute (KRS) 211.651-670, with definitions outlined in Kentucky Administrative Regulations (902 KAR 19:010).

The mission of KBSR is to promote early and accurate identification of children with birth anomalies and other disabling conditions in order to facilitate prevention, planning, and service delivery in the Commonwealth of Kentucky. KBSR achieves this mission through four primary areas.

- Surveillance - KBSR maintains a registry of birth defects in Kentucky, to allow for analysis of patterns and trends in time and geography. Data are aggregated, analyzed, and disseminated. Surveillance is the primary function of KBSR, which supports all other activities.
- Research - KBSR does not conduct research but facilitates studies by providing access to data.
- Prevention - KBSR partners with health education organizations to provide accurate information about the causes, impact, and prevention of birth defects.
- Services - KBSR identifies children at risk for developmental delays or disabilities and refers those families to the Kentucky Early Intervention System (KEIS). In 2024, KBSR identified 260 children for KEIS referrals.

Section 3: Methodology

Surveillance Methods

KBSR utilizes a web-based database to collect, link, and track information on children from birth to age five who are diagnosed with conditions defined in 902 KAR 19:010. KBSR is a passive surveillance system that currently receives data from the Kentucky Hospital Association (KHA), live birth certificates, death certificates, stillbirth certificates, Critical Congenital Heart Defect (CCHD) surveillance, outpatient clinics, and genetics laboratories. At earlier points in the period covered by this report, KBSR also incorporated elements from the U.S. Zika Pregnancy Registry, Zika Birth Defects Surveillance, and Kentucky's Public Health Neonatal Abstinence Syndrome Reporting Registry.

Inpatient data from KHA is considered the backbone of KBSR, with 96.9% of children with birth defects born in 2015-2023 having data from at least one admission. In this same group of children, 92.4% were linked to their live birth or stillbirth certificate, and 80.1% were linked to their CCHD screening. All other data sources reported for 0.5%-2.5% of cases. On average, birth defect cases in KBSR have information from 2.8 different reporting sources. The average time from birth to case creation was 239.2 days. Data from 2024 were excluded in these calculations as linkage may be incomplete. In the full period covered by this report, a total of 99,749 children were reported to KBSR, of which 72,114 (72.3%) were determined to have birth defects rather than other conditions covered by 902 KAR 19:010 (Figure 1).

To confirm records gathered by passive surveillance, nurses conduct medical chart reviews to verify selected diagnoses. Each code reviewed is marked either "Code confirmed" if the nurse confirms the diagnosis or "Code removed" if the nurse rules out the assigned code. The nurse then completes a narrative documenting the process. Since 2021, KBSR has performed this chart abstraction for all core, all recommended, and nearly all extended conditions defined by the National Birth Defects Prevention Network (NBDPN) (2004) in addition to other conditions of interest in the state. The goal is to complete abstractions by the child's second birthday. An epidemiologist performs a second-level clinical review on a random sample of approximately 10% of abstracted cases as a further quality check. The target date for clinical review is 30 days from abstraction, with an additional 10 days to correct any errors.

In 2024, a total of 1,866 KBSR cases had medical chart abstraction. The average time from birth to abstraction was 596 days. About half (49.8%) of these cases had at least one ICD code changed based on the medical chart review. Of the abstracted cases, 14.5% received second-level review, which occurred on average 42 days after abstraction. Of those receiving second-level review, 4.4% had a documentation error. All errors were corrected the same day they were noted.

Report Methods

All data elements relating to children with birth defects and their mothers were obtained from KBSR, although originally reported from a variety of data sources. The comparison group of all mothers giving birth in Kentucky was obtained from the certificates of live birth.

Dataset inclusion criteria

This report covers birth defects data spanning birth years 2015 to 2024, reported as of Summer 2025. Cases were restricted to Kentucky residents based on the mother's state of residence, or when missing, the child's state of residence. Data from individual years were appended together to create a cohesive ten-year dataset. In this report, the term "birth defects" is limited to include only conditions both in regulation and contained within the range of International Classification of Diseases (ICD) codes 740.0 to 759.9 (for ICD-9) or within the Q range (ICD-10). Any ICD code that was ruled out during medical chart review was excluded.

Definitions

When possible, definitions were aligned with NDBPN or CDC; however, differences in methodology may limit comparability with other reports. Major birth defects are defined as those that are core, recommended, or extended birth defects by the NBDPN definition. Defects are grouped into nine categories, though not all birth defects fall into a category. Children may have multiple birth defects in one category, but the report deduplicates cases so that the total number of children per category is represented in the dataset. Fatalities include any documented fetal or infant death recorded on vital records or hospital discharge data.

Analytic methods

Cases are unduplicated, and each case represents one individual child, infant, or fetus with the specified birth defect (defined by ICD codes). Rate calculations are the estimated prevalence of conditions per 10,000 live births. Although the numerator includes both stillbirths and live births, only live births are used in the denominator in accordance with standard practices of the CDC and other entities reporting birth defects. Condition-specific fatality rate is the percentage of cases with a condition that ended in stillbirth or death.

Rates are suppressed if a cell count is <5 to prevent the identification of an individual case. If the cell's original size can be determined by subtraction from the total, then the exact number of the next smallest cell is also suppressed. Rates that are based on cells with counts ranging from 5 to 19 are noted as being unstable. The numbers are very small, represent rare conditions, and should be interpreted with caution. At times, 3-year or 5-year rolling averages are used to increase the case counts per data point, which can improve the stability of the estimates and reduce the impact of outliers. When data were analyzed by geography, the unit of aggregation is the Area Development District (ADD), as a multi-county region is less likely to result in data suppression or identification of individual cases.

Data compilation and analyses were performed using SAS, version 9.4, and mapping was performed using Esri ArcGIS Pro.

Limitations

The primary limitation of this report is that the KBSR uses passive surveillance. Variation in external reporting sources can affect the completeness and timeliness of data reporting. Although children may be reported up to age five, more recent birth years (particularly 2020 through 2024) should be considered preliminary due to reporting lag.

Variation in diagnostic practices across facilities and clinicians may also influence reported prevalence. While standardized medical chart abstraction helps to reduce misclassification, differences in clinical evaluation, access to specialty care, and coding practices can affect case identification. Additionally, Kentucky statute does not require reporting from many outpatient facilities or from out-of-state providers. As a result, some Kentucky residents who receive care in neighboring states may not be captured in the registry, potentially leading to underestimation in border regions.

A final limitation of the data in this report is that over the course of a 10-year period, there have been numerous programmatic and methodology changes within the Kentucky Birth Surveillance Registry. There have been changes in the conditions abstracted, the switch from ICD-9 to ICD-10 coding, data sources included in the registry, and areas of specific focus and interest within the program. It is difficult to assess the impact that these methodological changes have on the data in the report, but using multi-year rolling averages makes the rates more comparable.

Section 4: Prevalence of Selected Birth Defects

There were 72,114 children with birth defects reported to KBSR in this 10-year period, ranging from 6,153 to 8,520 per year. Reporting for 2024 was approximately 20% lower than previous years; that reduction is likely due to the reporting timelines discussed in the methodology.

When limiting the dataset to major birth defects, as described in the methodology, there were 20,235 children, equal to a rate of 380 cases per 10,000 live births. Approximately every 1 in 26 children in Kentucky has a major birth defect, which is about 25% higher than the national estimates (CDC, 2025). The rate of major birth defects per year is visualized in Figure 2.

There are a few geographic patterns worth noting. First is that the highest rates of birth defects are in eastern Kentucky, in the Kentucky River and Big Sandy Area Development Districts (ADDs) (590.50 and 527.22 per 10,000 live births, respectively). The lowest rates of birth defects are in Northern Kentucky, Buffalo Trace, and Pennyrile ADDs (140.33, 210.37, and 211.08 per 10,000 live births, respectively). All three of these ADDs are in close proximity to major metropolitan areas and specialty children's hospitals, which could lead to lower reporting among Kentucky residents seeking out-of-state medical care. The rate of birth defects by ADD is visualized in Figure 3.

Figure 4 presents the five-year rolling average rates of major birth defect categories per 10,000 live births from 2015 to 2024. Cardiovascular defects consistently had the highest prevalence during this study period, although rates show a gradual decline over time. Genitourinary defects increased steadily through 2023 before slightly leveling in the most recent five-year period, while other defects remained relatively stable throughout the 10-year period. Overall, the rolling averages suggest relative stability across most categories, with notable decreases in cardiovascular defects.

Table 1 and Table 2 present counts and prevalence rates of major birth defects identified by the KBSR from 2015 to 2024. Table 1 provides overall rates to illustrate trends over time, while Table 2 displays 5-year rolling averages to illustrate trends over time. Rates are reported per 10,000 live births, with suppression applied when counts are small.

Central Nervous System Anomalies

There was an average of 254 children per year with any central nervous system (CNS) anomaly, equal to a rate of 47.69 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 5.

Neural tube defects

NTDs are a group of CNS anomalies that occur when the neural tube fails to close in the first few weeks of fetal development; they can affect the brain, skull, spinal cord, and vertebrae (CDC, 2025b). Three NTDs are considered major birth defects in this report: anencephaly, spina bifida, and encephalocele. An estimated 32 children per year in Kentucky have any kind of NTD. That equates to a rate of 5.95 per 10,000 live births (Figure 6), which is a 21.4% increase from the rate of 4.9 included in KBSR's 2005-2014 report (Kentucky Department for Public Health [KDPH], 2016).

Eye Anomalies

Eye anomalies are the rarest category of birth defects discussed in this report. There was an average of 33 children per year with any eye anomaly, equal to a rate of 6.12 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 7.

Ear, Face, and Neck Anomalies

There was an average of 168 children per year with any anomaly of the ear, face, or neck, equal to a rate of 31.63 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 8.

Cardiovascular Anomalies

Cardiovascular anomalies are the most common category of birth defects discussed in this report. There was an average of 2,041 children per year with any cardiovascular anomaly, equal to a rate of 383.34 per 10,000 live births. That rate decreased slightly across the period of 2015-2024 and is visualized in Figure 9.

Critical Congenital Heart Defects (CCHDs)

By the authority of KRS 214.155, Critical Congenital Heart Defect (CCHD) pulse oximetry screening is conducted for newborns in Kentucky prior to hospital discharge. This screening helps identify infants with some serious heart defects who may not show visible symptoms at birth. Early detection through CCHD screening improves clinical outcomes and reduces risk of morbidity and mortality associated with delayed diagnosis.

Conditions commonly designated as CCHDs or "primary lesions" include hypoplastic left heart syndrome, dextro-transposition of the great arteries, pulmonary atresia, truncus arteriosus, total anomalous pulmonary venous return, tricuspid atresia, and tetralogy of Fallot. Conditions designated in Kentucky as "secondary lesions" that are also CCHDs include aortic interruption/atresia/hypoplasia, coarctation/hypoplasia of aortic arch, double-outlet right

ventricle, Ebstein anomaly, and single ventricle. The rates of primary and secondary lesions are visualized in Figure 10.

Orofacial Anomalies

There was an average of 212 children per year with any respiratory or orofacial anomaly, equal to a rate of 39.89 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 11. Choanal atresia is grouped with orofacial anomalies in this report, although it is sometimes considered an anomaly of the respiratory system. Orofacial clefts include cleft palate and cleft lip, which can occur in isolation or together.

Gastrointestinal Anomalies

There was an average of 1,285 children per year with any gastrointestinal anomaly, equal to a rate of 241.29 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 12.

Genitourinary Anomalies

There was an average of 1,169 children per year born with any genital or urinary anomaly, equal to a rate of 241.29 per 10,000 live births. That rate increased slightly across the period of 2015-2024 and is visualized in Figure 13.

Musculoskeletal Anomalies

There was an average of 568 children per year born with any muscular system or skeletal anomaly, equal to a rate of 106.72 per 10,000 live births. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 14. One noteworthy defect in this category is gastroschisis, which is a defect of the abdominal wall. The prevalence of gastroschisis has been increasing in the U.S. in recent decades (Jones et al., 2016; Short et al., 2019); no increase was found in this sample, but the estimated prevalence in this sample is about 10 times higher than it was in 2005-2007 (KDPH, 2016).

Chromosomal Anomalies

There was an average of 181 children per year born with any chromosomal anomaly, equal to a rate of 33.98 per 10,000 live births. This category includes conditions such as Trisomy 21, Turner Syndrome, and many others that affect specific chromosomes. That rate remained fairly consistent across the period of 2015-2024 and is visualized in Figure 15.

Section 5: Maternal Risk Factors

Maternal demographics and health concerns provide an underlying context for the study of birth defects (CDC, 2025). Identifying at-risk groups could allow for the development of targeted interventions for birth defects education and prevention.

Full results for maternal health factors are in Table 3. Of note, "unknown" values were high for children with birth defects across all categories (7.4%-17.0%), as not all KBSR cases are linked to the certificate of live birth. For that reason, in this section and in the data tables, unknown values have been excluded, and other categories were adjusted accordingly.

Maternal Age

Gastroschisis is more common when the mother is young, especially in the teenage years and early twenties (CDC, 20205; Jones et al., 2016). In this dataset, the highest rate of gastroschisis is among children born to mothers <20 years old (12.76 per 10,000 live births), followed by mothers 20-24 years old (6.77 per 10,000 live births). The rate continues dropping among children of mothers 25-29 years old (3.21 per 10,000 live births) and is lowest among those 30 years or older. Rates by age are visualized in Figure 16.

Older mothers are more likely to have children with Trisomy 21, especially when maternal age is above 35 years (CDC, 2025). In this dataset, the highest rate of Trisomy 21 is among children born to mothers who are 40 or more years old (85.48 per 10,000 live births), followed by mothers 35-39 years old (29.19 per 10,000 live births). The rates are lower among children of mothers who are less than 35 years old. Rates by age are visualized in Figure 17.

Diabetes

Maternal diabetes is a well-established risk factor for several major birth defects, including congenital heart defects and orofacial clefts (CDC, 2025c; CDC, 2026). In the analysis, pre-existing diabetes was more common among mothers in KBSR compared to the general population of mothers giving birth in Kentucky (Figure 18). However, because there were few substantial differences in birth defect prevalence between mothers with gestational diabetes and those without diabetes, this section focuses on pre-existing diabetes only.

Compared to mothers without diabetes, the odds of several conditions were elevated among mothers with pre-existing diabetes. The most substantially increased odds ratios (OR) were for renal agenesis/hypoplasia (OR: 6.73), any NTD (OR: 4.20), any heart defect (OR: 4.07), clubfoot (OR: 2.99), any orofacial cleft (OR: 2.49), and hypospadias (OR: 1.40). These findings are visualized in Figure 19.

Hypertension

Pre-existing hypertension was more common among mothers in KBSR, as shown in Figure 18. Compared to mothers without hypertension, the odds of several conditions were elevated among mothers with pre-existing hypertension. The most substantially increased odds ratios (OR) were for large intestinal atresia (OR: 2.44), renal agenesis/hypoplasia (OR: 1.74), any heart defect (OR: 1.59), and any orofacial cleft (OR: 1.45). These findings are visualized in Figure 20. Overall, these odds ratios are more modest than the impact of pre-existing diabetes.

Pre-pregnancy Weight

When examining body mass index (BMI) category, most mothers giving birth were overweight or obese (59.8% among mothers of children with birth defects, 58.6% among all Kentucky mothers). In both groups, about one in three had a normal pre-pregnancy weight, and few mothers were underweight. Pre-pregnancy obesity was associated with slightly increased odds of any orofacial cleft (OR: 1.32), any heart defect (OR: 1.28), and any NTD (OR: 1.27). The rate of gastroschisis was highest for mothers with underweight BMIs (10.18 per 10,000 live births) and dropped as BMI increased (Figure 21).

Smoking

Non-smoking mothers represented the largest percent of births both among mothers of children with birth defects and among all Kentucky births (86.7% vs. 87.6%, respectively). Heavy smokers (smoking greater than 10 cigarettes per day in the third trimester) were relatively uncommon both among mothers of children with birth defects (4.0%) and among all Kentucky births (3.7%). Non-heavy smoking (less than 10 cigarettes per day in the third trimester) was more common than heavy smoking among both mothers of children with birth defects (9.3%) and among all Kentucky births (8.7%). Smoking during pregnancy was associated with increased odds of gastroschisis (OR: 2.55), choanal atresia (OR: 1.99), any orofacial cleft (OR: 1.74), and any limb reduction defect (OR: 1.60). These findings are visualized in Figure 22.

Heart defects were more common among mothers with heavy smoking during pregnancy (433.11 per 10,000 live births), although non-heavy smokers still had slightly elevated rates compared to non-smokers (395.62 and 331.18 per 10,000 live births, respectively). The data are not included in a table but are shown in Figure 23.

Section 6: Pregnancy Outcomes

Preterm Birth

Most births occurred at or beyond 37 weeks of completed gestation, making them term pregnancies (80.0% among KBSR, 88.6% among all Kentucky births). Full term (39+ weeks) was more common than early term (37-38 weeks) among both samples. Births occurring before 37 weeks of completed gestation, broadly considered premature or preterm, were more common among children with birth defects (20.1%) than Kentucky births overall (11.4%). The difference was especially pronounced as gestational age decreased: late preterm (11.4% and 8.4%, respectively), preterm (2.9% and 1.3%, respectively), and early preterm (5.8% and 1.7%, respectively). Categories of preterm birth are visualized in Figure 24, and gestational age data are displayed in full in Table 4.

Preterm infants are more likely to have heart defects, and infants with heart defects are more likely to be preterm (Reddy et al., 2022). Some heart defects among preterm infants may resolve naturally as the child develops, but others require intervention. In this dataset, the rate of cardiovascular anomalies decreased as gestational age increased, from 3,225.37 per 10,000 live births among those born at <32 weeks gestation down to 203.78 per 10,000 live births among those born at 39 or more weeks gestation. Approximately one out of every three children born at <32 weeks of gestation is diagnosed with any cardiovascular anomaly. The rates of heart defects among all categories of gestational age are visualized in Figure 25.

Birth Defects Mortality

Birth defects are the leading cause of infant deaths before the first birthday in the U.S (CDC, 2025). In Kentucky, birth defects are the second most common cause of infant death and the most common cause of death before the eighteenth birthday, with about 74 deaths per year (Kentucky Cabinet for Health & Family Services [CHFS], 2025). In both Kentucky and the U.S., birth defects are responsible for about 1 in every 5 infant deaths (CDC, 2025; CHFS, 2025).

In this sample, there were 1,665 children whose mortality was documented in KBSR. There were 1,308 deaths and 357 stillbirths (Table 5). Most deaths (90.1%) and all stillbirths are linked to their vital records. For those with death certificate data, the most common manner of death was natural (85.1%). The manner of death could not be determined in 8.6% of deaths, and 4.9% were accidental deaths. About a third (30.3%) of those with a death certificate had a birth defect listed as the primary cause of death. The most common type of birth defect listed as the primary cause of death was CNS anomalies (97 deaths), especially anencephaly, followed by chromosomal anomalies (77 deaths), especially Trisomy 13 and Trisomy 18, and heart anomalies (76 deaths). Most children in this dataset with a death certificate (84.2%) died during the first year of life.

KRS 213.096 establishes that stillbirth certificates are given in the event of a spontaneous fetal death of twenty weeks or more gestation or in which the fetus weighs 350 grams or more. Among stillbirths in the sample, 41.5% had a birth defect listed as the initiating condition. The most common type of birth defect listed was chromosomal anomalies (54 stillbirths), followed by CNS anomalies (36 stillbirths). The most common types of birth defects listed as the primary cause of death or the initiating cause of stillbirth are in Table 6 and Figure 26.

Overall, CNS anomalies were the most common type of birth defect to cause death or stillbirth in this sample (133 cases), with anencephaly being the most common individual condition. There is a strong connection between heart defects and prematurity (Reddy et al., 2022), and they are both drivers of infant mortality (CDC, 2024; CHFS, 2025). Overall, cardiovascular anomalies were the third most common type of birth defect to cause death or stillbirth in this sample (89 cases). Anomalies of the kidneys and bladder caused 34 deaths or stillbirths in this sample. Musculoskeletal and limb anomalies caused 38 deaths or stillbirths in this sample. Overall, chromosomal anomalies were the second most common type of birth defect to cause death or stillbirth in this sample (131 cases).

Fatality Rates for Selected Birth Defects

Some birth defects have high fatality rates, meaning that they are likely to end in death or stillbirth. Birth defects with fatality rates exceeding 25% in this sample are shown in Figure 27.

Anencephaly is considered a lethal anomaly (CDC, 2026b), and in this sample, 96.3% of anencephaly cases resulted in mortality. Encephalocele is another serious NTD that is likely to end in mortality (62.0% of cases in this report). Holoprosencephaly is not an NTD but is a different type of CNS anomaly, and 76.5% of cases in this sample resulted in mortality.

The heart defect with the highest mortality rates was hypoplastic left heart syndrome (46.7%), followed by Ebstein anomaly (33.3%), single ventricle (28.8%), double-outlet right ventricle (36.1%), and endocardial cushion defects (28.0%). Of these, all but endocardial cushion defects are primary or secondary lesions that have the potential for detection by CCHD screening, providing opportunities for early intervention.

Two musculoskeletal defects have high mortality rates: diaphragmatic hernia (31.1%) and omphalocele (38.2%), which is an abdominal wall defect.

Trisomy 13 and Trisomy 18 are autosomal trisomy disorders, and they are associated with poor pregnancy outcomes. The median first-year mortality among live births with Trisomy 13 and Trisomy 18 is 87% and 88%, respectively (Goel et al., 2019). In this sample, 85.9% of cases of Trisomy 13 and 85.8% of cases of Trisomy 18 resulted in mortality, and those numbers may reflect under-reporting of deaths.

Section 7: Public Health Implications

About 1 in every 33 births in the United States (U.S.) is affected by a major birth defect that requires medical intervention or has serious impacts on health or development (CDC, 2025; NBDPN, 2004); this report estimates that in Kentucky, the figure is closer to 1 in every 26. When considering all birth defects, not just the major birth defects that require medical intervention or have serious impacts on the child, there are more than 7,200 new births affected every year in Kentucky, meaning that birth defects are an extremely common public health concern.

In addition to being common, some birth defects are very serious; in both Kentucky and the U.S., birth defects are responsible for about 1 in every 5 infant deaths and are a major driver of mortality among babies and children (CDC, 2025; CHFS, 2025). Although not all birth defects are preventable, supporting healthy pregnancies promotes positive pregnancy outcomes, and having updated Kentucky-specific data is the first step in tailoring prevention strategies.

Because of the extensive quality checks that are part of KBSR protocols, the dataset is often used to support academic research and governmental evaluations. During the past 10 years, KBSR has been featured in four peer-reviewed studies that utilized data from birth defects surveillance programs across the nation. Two were about the increasing prevalence of gastroschisis (Jones et al., 2016), an abdominal wall defect, and the possible link to opioid exposure (Short et al., 2019), and two were about congenital Zika virus and Zika-associated birth defects (Roth et al., 2022; Neelam et al., 2024). KBSR routinely participates in further national data collection as part of an initiative between NBDPN and the CDC to publish birth defects prevalence estimates, with the most recent publication in 2024 (Stallings et al., 2024). KBSR also partners with Kentucky Environmental Public Health Tracking in order to make county-level birth defects data publicly accessible (CDC, n.d.).

In the next 10 years, KBSR has opportunities to continue collaborating with current partners, to improve current processes, and to face emerging public health concerns. This report is just the first step in sharing birth defects information with partners in academia, medicine, public health, parent advocacy, and family spaces.

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Appendices

Appendix 1. Data Tables

Table 1. Rates of Major Birth Defects, page 1/2

	KY, Average children per year	KY, 2015-2024	KY, 2005-2014	U.S., 2016-2020
CENTRAL NERVOUS SYSTEM				
Anencephaly	11	2.01	1.6	2.28
Spina bifida	17	3.12	3.0	3.65
Encephalocele	5	0.94	NA	1.04
Holoprosencephaly	5	0.96	NA	NA
EYE				
Anophthalmia/microphthalmia	<5	0.81	NA	2.08
Congenital cataract	5	0.98	NA	NA
EAR, FACE, NECK				
Microtia	7	1.22	NA	NA
Anotia	<5	0.09	NA	NA
CARDIOVASCULAR				
Truncus	<5	0.60	NA	0.65
Transposition of the great arteries	18	3.31	NA	2.97
Tetralogy of Fallot	21	3.96	NA	4.89
Ventricular septal defect	358	67.28	NA	NA
Atrial septal defect	1,354	254.29	NA	NA
Endocardial cushion defects	19	3.62	NA	5.83
Pulmonary valve atresia and stenosis	28	5.24	NA	10.41
Tricuspid valve atresia and stenosis	<5	0.77	NA	1.83
Ebstein anomaly	<5	0.62	NA	0.81
Aortic valve stenosis	6	1.13	NA	NA
Hypoplastic left heart syndrome	12	2.25	NA	2.59
Coarctation of the aorta	28	5.24	NA	5.80
Total anomalous pulmonary venous connection	6	1.11	NA	1.38
Common ventricle	6	1.11	NA	0.68
Interrupted aortic arch	<5	0.45	NA	0.98
Double outlet right ventricle	14	2.70	NA	2.44

Table 1. Page 2/2

	KY, Average children per year	KY, 2015-2024	KY, 2005-2014	U.S., 2016-2020
OROFACIAL				
Choanal atresia	6	1.09	NA	NA
Cleft palate alone	36	6.67	NA	6.26
Cleft lip alone	18	3.40	NA	3.41
Cleft lip and palate	35	6.57	NA	6.54
GASTROINTESTINAL				
Esophageal atresia/tracheoesophageal fistula	11	2.01	NA	2.36
Rectal and large intestinal atresia/stenosis	24	4.47	NA	4.59
Biliary atresia	<5	0.83	NA	NA
Small intestinal atresia	16	3.04	NA	NA
GENITOURINARY				
Hypospadias**	195	71.61	NA	NA
Renal agenesis/hypoplasia	34	6.40	NA	NA
Bladder exstrophy	<5	0.13	NA	NA
Posterior urethral valves**	5	1.80	NA	NA
Cloacal exstrophy	<5	0.09	NA	NA
MUSCULOSKELETAL				
Diaphragmatic hernia	15	2.84	2.9	3.17
Omphalocele	11	2.07	1.1	2.63
Gastroschisis	22	4.15	2.6	2.36
Reduction deformities	20	3.76	NA	5.07
Clubfoot	102	19.16	NA	18.87
CHROMOSOMAL				
Trisomy 21	68	12.83	11.6	16.33
Trisomy 13	7	1.33	NA	1.55
Trisomy 18	13	2.52	1.5	3.27
Turner syndrome^	42	16.31	NA	NA

*Rates based on counts <20.

**Rate per 10,000 *male* live births

^Rate per 10,000 *female* live births

Table 2. Rates of Major Birth Defects in Kentucky, page 1/2

	2015-2019	2016-2020	2017-2021	2018-2022	2019-2023	2020-2024
CENTRAL NERVOUS SYSTEM						
Anencephaly	2.06	2.21	2.32	2.39	1.99	1.88
Spina bifida	3.72	3.43	3.39	3.28	2.84	2.39
Encephalocele	0.92	0.92	0.85	0.88	0.88	0.92
Holoprosencephaly	1.14	1.03	1.07	0.96	0.74	0.74
EYE						
Anophthalmia/microphthalmia	0.99	0.88	0.85	0.63*	0.48*	0.59*
Congenital cataract	1.22	1.03	0.96	0.85	0.88	0.70*
EAR, FACE, NECK						
Microtia	0.92	1.14	1.14	1.36	1.66	1.47
Anotia	***	***	***	***	***	***
CARDIOVASCULAR						
Truncus	0.66*	0.66*	0.48*	0.48*	0.48*	0.52*
Transposition of the great arteries	3.35	3.13	3.61	3.46	3.24	3.13
Tetralogy of Fallot	4.05	3.39	3.24	3.35	3.61	3.72
Ventricular septal defect	65.85	63.64	63.86	64.19	67.58	66.07
Atrial septal defect	336.13	327.51	318.20	297.68	230.77	162.49
Endocardial cushion defects	4.31	4.05	4.01	3.79	3.50	2.80
Pulmonary valve atresia and stenosis	4.71	5.12	5.56	5.93	6.00	5.56
Tricuspid valve atresia and stenosis	0.81	0.59*	0.63*	0.70*	0.55*	0.70*
Ebstein anomaly	0.70*	0.55*	0.63*	0.63*	0.66*	0.52*
Aortic valve stenosis	0.96	0.92	0.88	0.88	1.03	1.25
Hypoplastic left heart syndrome	2.10	2.28	2.43	2.50	2.50	2.32
Coarctation of the aorta	5.41	4.79	4.60	4.46	4.35	4.86
Total anomalous pulmonary valve return	1.25	1.14	1.07	1.03	0.88	0.92
Common ventricle	1.07	0.92	1.14	1.10	0.99	1.10
Interrupted aortic arch	0.26*	0.37*	0.63*	0.55*	0.59*	0.63*
Double outlet right ventricle	2.06	2.54	2.87	2.73	3.13	3.24

Table 2. Page 2/2

	2015-2019	2016-2020	2017-2021	2018-2022	2019-2023	2020-2024
OROFACIAL						
Choanal atresia	1.40	1.33	1.40	1.10	0.99	0.74
Cleft palate alone	6.96	6.67	6.56	6.74	6.44	6.11
Cleft lip alone	3.57	2.95	2.91	3.06	3.09	3.09
Cleft lip and palate	6.41	6.22	6.44	6.04	6.56	6.48
GASTROINTESTINAL						
Esophageal atresia/TEF	1.95	2.25	2.14	2.10	2.03	1.99
Rectal/ large intestinal atresia/stenosis	5.16	4.75	4.38	4.16	3.98	3.61
Biliary atresia	0.96	0.92	0.96	0.85	0.66*	0.66*
Small intestinal atresia	3.20	2.80	2.94	3.13	2.91	2.76
GENITOURINARY						
Hypospadias**	75.23	82.78	81.91	79.0	74.73	64.96
Renal agenesis/hypoplasia	5.49	5.52	5.78	6.48	6.81	7.07
Bladder exstrophy	***	***	***	***	***	***
Posterior urethral valves**	1.51	1.65	1.58	1.58	1.65	2.01
Cloacal exstrophy	***	***	***	***	***	***
MUSCULOSKELETAL						
Diaphragmatic hernia	2.65	2.39	2.76	3.20	3.13	2.91
Omphalocele	2.17	2.14	2.47	2.43	2.14	1.88
Gastroschisis	4.35	4.12	4.35	4.53	4.20	3.79
Reduction deformities	3.76	3.65	3.76	3.87	3.72	3.61
Clubfoot	20.96	19.57	19.00	18.78	17.46	16.61
CHROMOSOMAL						
Trisomy 21	13.37	12.01	11.75	12.04	12.56	11.79
Trisomy 13	1.25	1.10	1.58	1.47	1.33	1.36
Trisomy 18	2.69	2.54	2.54	2.39	2.28	2.25
Turner syndrome^	13.08	15.80	17.84	18.37	18.83	18.98
GENITOURINARY						
Hypospadias**	75.23	82.78	81.91	79.0	74.73	64.96
Renal agenesis/hypoplasia	5.49	5.52	5.78	6.48	6.81	7.07
Bladder exstrophy	***	***	***	***	***	***
Posterior urethral valves**	1.51	1.65	1.58	1.58	1.65	2.01

*Rates based on counts <20.

**Rate per 10,000 *male* live births

^Rate per 10,000 *female* live births

***Data suppressed due to counts <5.

Table 3. Maternal Health Factors by KBSR Cases and Kentucky Live Births, 2015-2024

		KBSR N=99,749	KENTUCKY N=532,427
MATERNAL AGE	<20	6.6	6.7
	20-24	25.2	25.2
	25-29	30.4	30.8
	30-34	24.0	24.2
	35-39	11.2	10.6
	40+	2.6	2.5
DIABETES	Pre-existing	2.0	1.2
	Gestational	7.2	7.5
	None	90.7	91.3
HYPERTENSION	Pre-existing	3.6	3.2
	Gestational	10.0	10.0
	None	86.4	86.8
WEIGHT	Underweight	3.7	3.5
	Normal	36.6	37.9
	Overweight	25.2	25.7
	Obese	34.6	32.9
SMOKING	Heavy Smoker	4.0	3.7
	Non-Heavy Smoker	9.3	8.7
	Non-Smoker	86.7	87.6
PRENATAL CARE	Above Standard	20.0	18.2
	Standard	33.9	35.6
	Below Standard	43.9	44.5
	No Visits	2.2	1.7
GESTATIONAL AGE	Early Preterm (<32 weeks)	5.8	1.7
	Preterm (32-33 weeks)	2.9	1.3
	Late Preterm (34-36 weeks)	11.4	8.4
	Early Term (37-38 weeks)	30.9	31.0
	Full Term (39+ weeks)	49.1	57.6

Table 4. Gestational Age of Livebirths by KBSR Cases and Kentucky Live Births, 2015-2024

		KBSR N=99,749	KENTUCKY N=532,427
GESTATIONAL AGE	Early Preterm (<32 weeks)	5.8	1.7
	Preterm (32-33 weeks)	2.9	1.3
	Late Preterm (34-36 weeks)	11.4	8.4
	Early Term (37-38 weeks)	30.9	31.0
	Full Term (39+ weeks)	49.1	57.6

Table 5. Documented Mortality Among Children with Birth Defects

	DOCUMENTED MORTALITY N=1665	
	n	%
Stillbirth	357	
Stillbirth certificate	357	100.0%
Other documentation of stillbirth	0	0.0%
Death	1308	
Death certificate	1180	90.1%
Other documentation of death	129	9.9%
Manner of death		
Accident	58	4.9%
Could not be determined	102	8.6%
Natural	1004	85.1%
All other manners of death	16	1.4%

Table 6. Count of Causes of Death or Stillbirth, by Category of Birth Defect

	TOTAL (N=505)	DEATH (N=357)	STILLBIRTH (N=148)
Central nervous system anomalies	133	97	36
Chromosomal anomalies	131	77	54
Heart defects	89	76	13
Other anomalies	81	61	20
Musculoskeletal and limb anomalies	38	24	14
Lung hypoplasia	37	37	0
Anomalies of the kidney and bladder	34	23	11

Appendix 2. Figures

Figure 1. Number of Children Reported to KBSR by Year

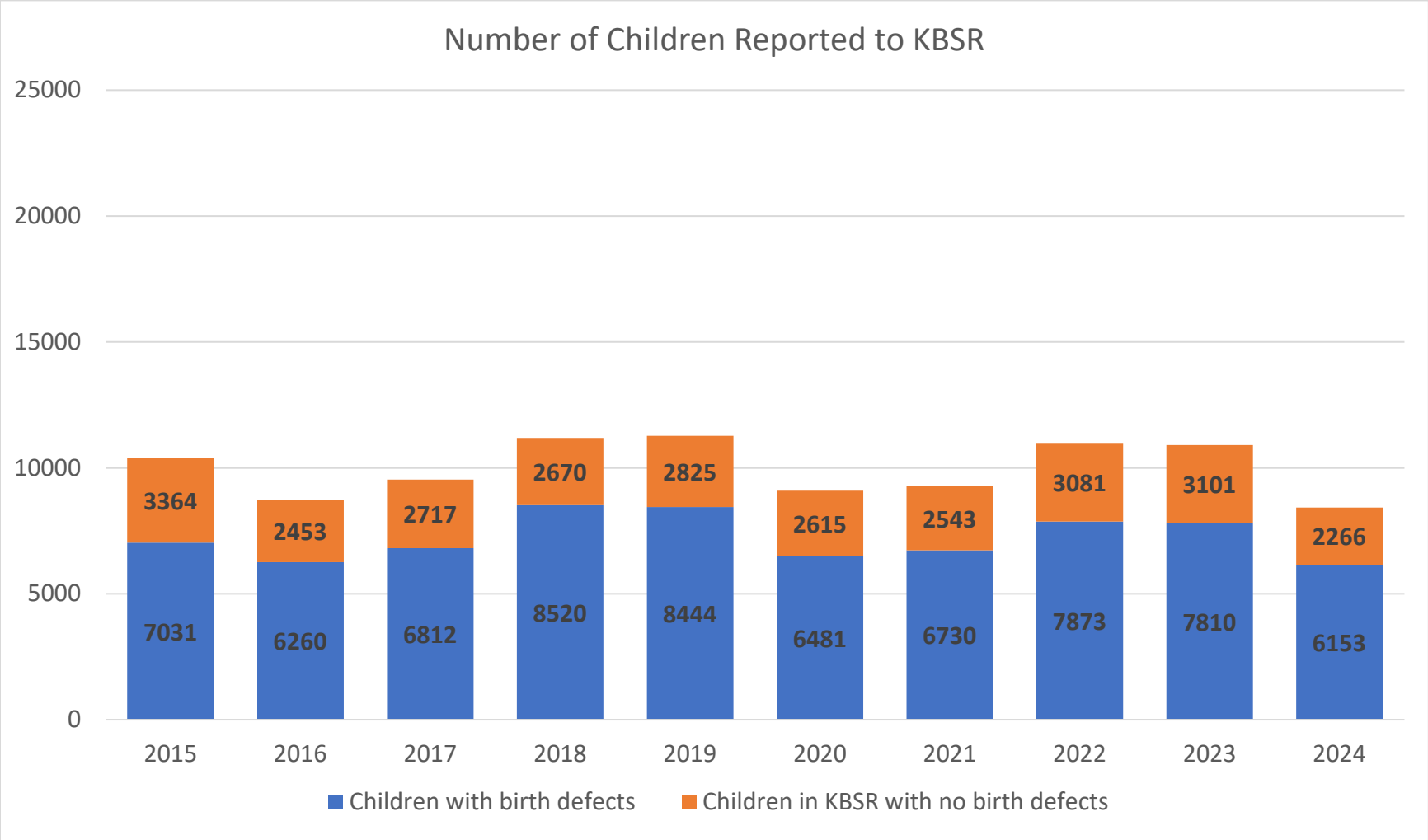

Figure 2. Rate of Major Birth Defects, 2015-2024

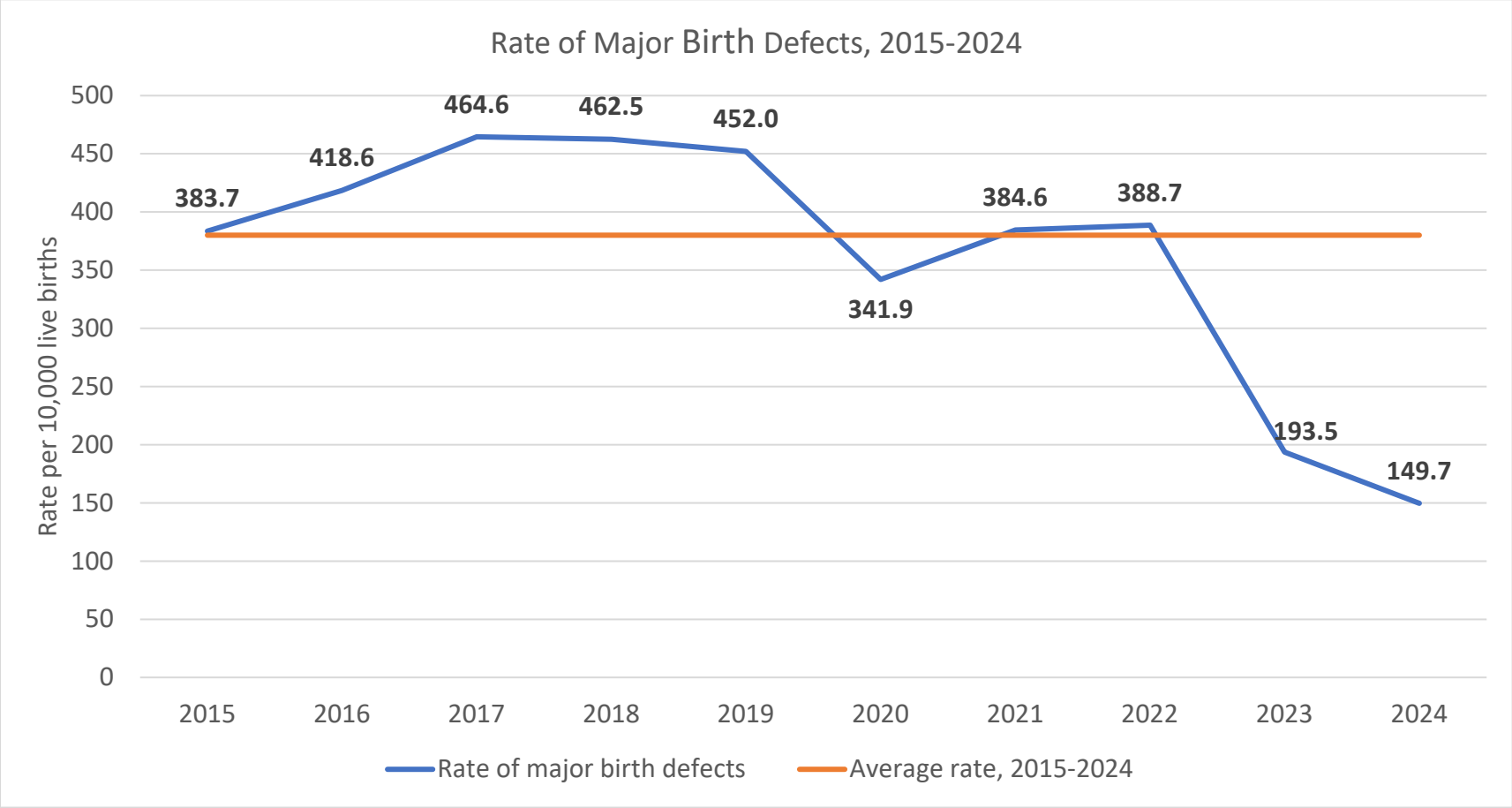

Figure 3. Rate of Major Birth Defects by Area Development District, 2015-2024

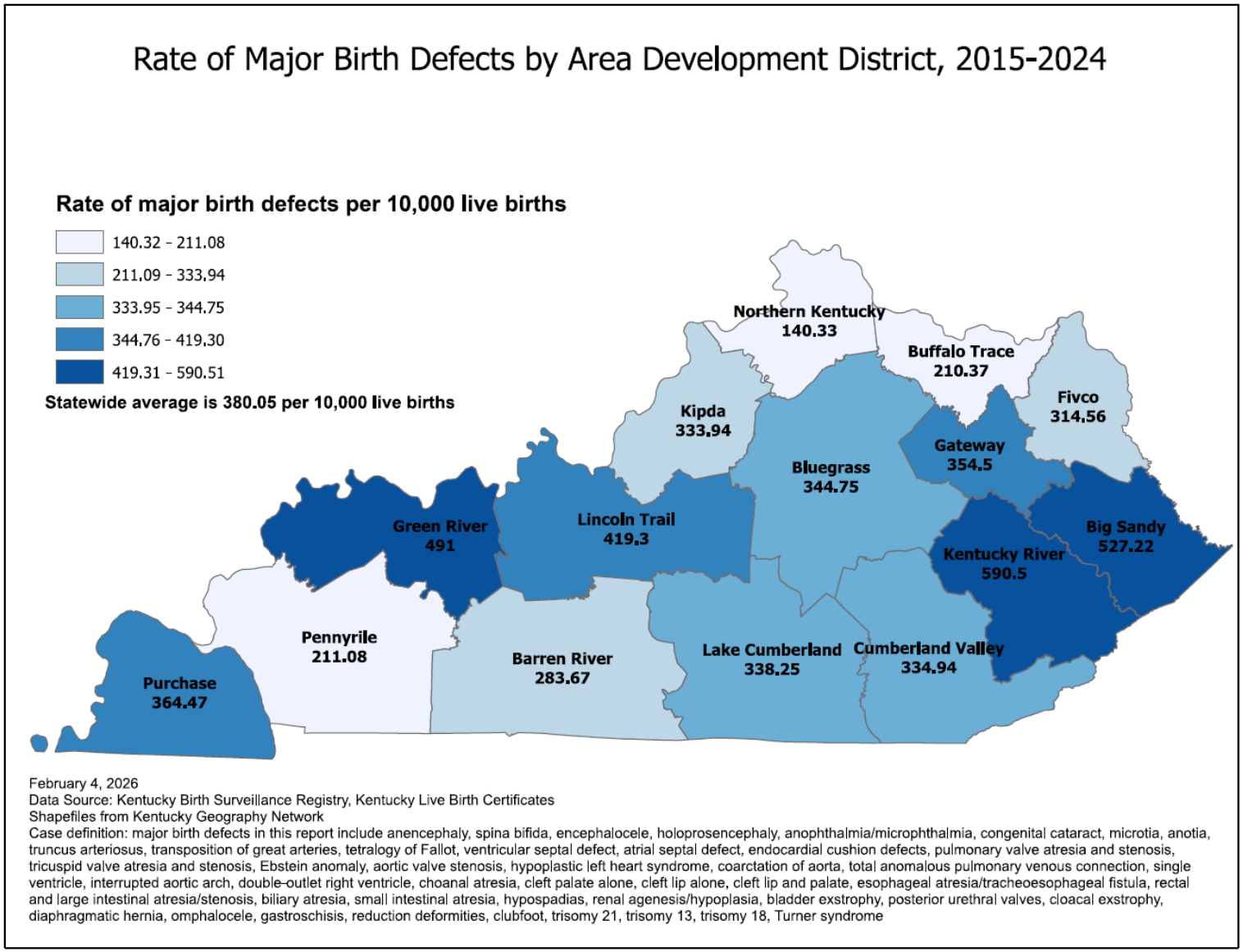

Figure 4. 5-Year Rolling Average Rate of Birth Defects Categories

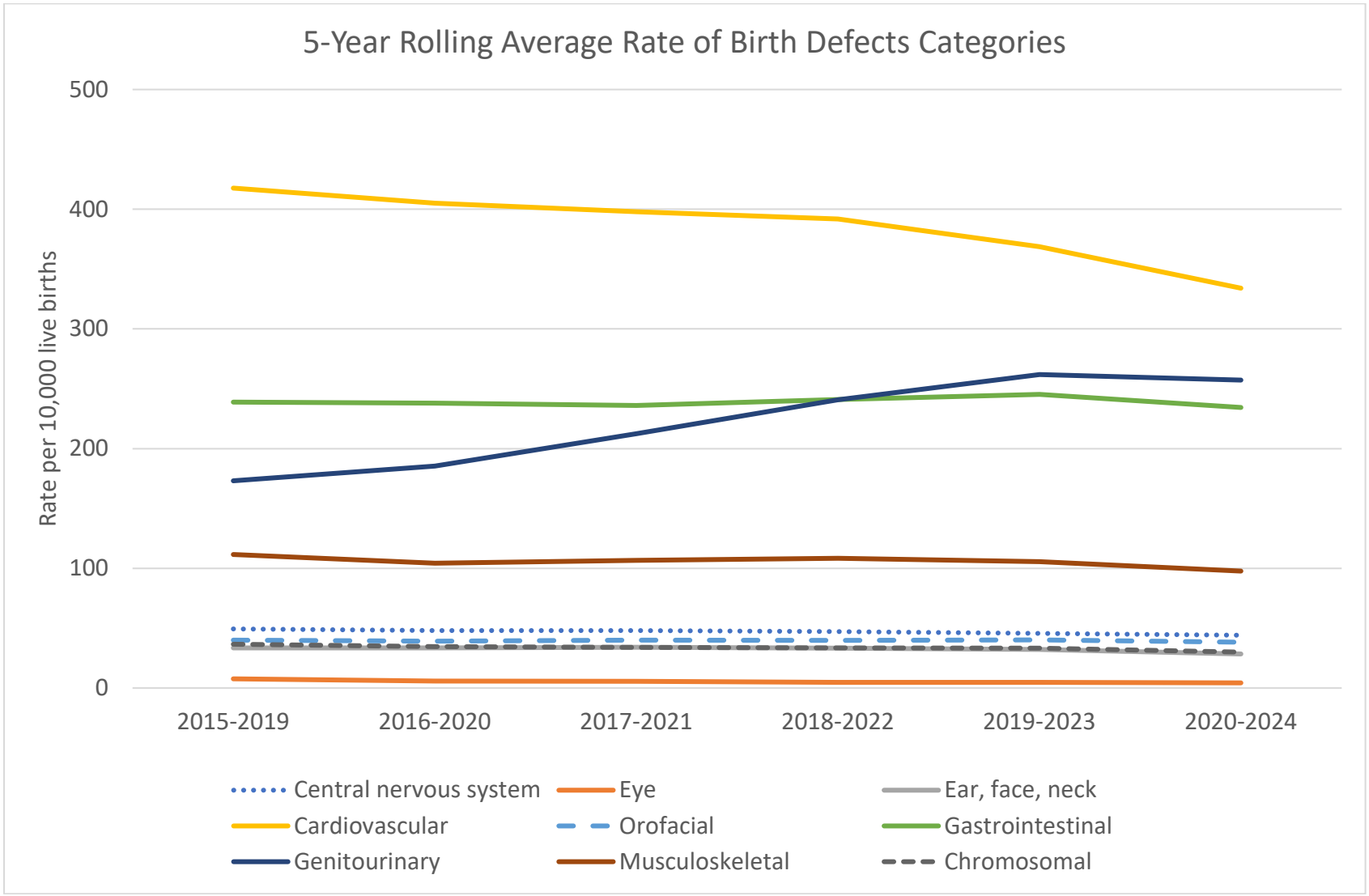

Figure 5. 3-Year Rolling Average Rate of CNS Anomalies

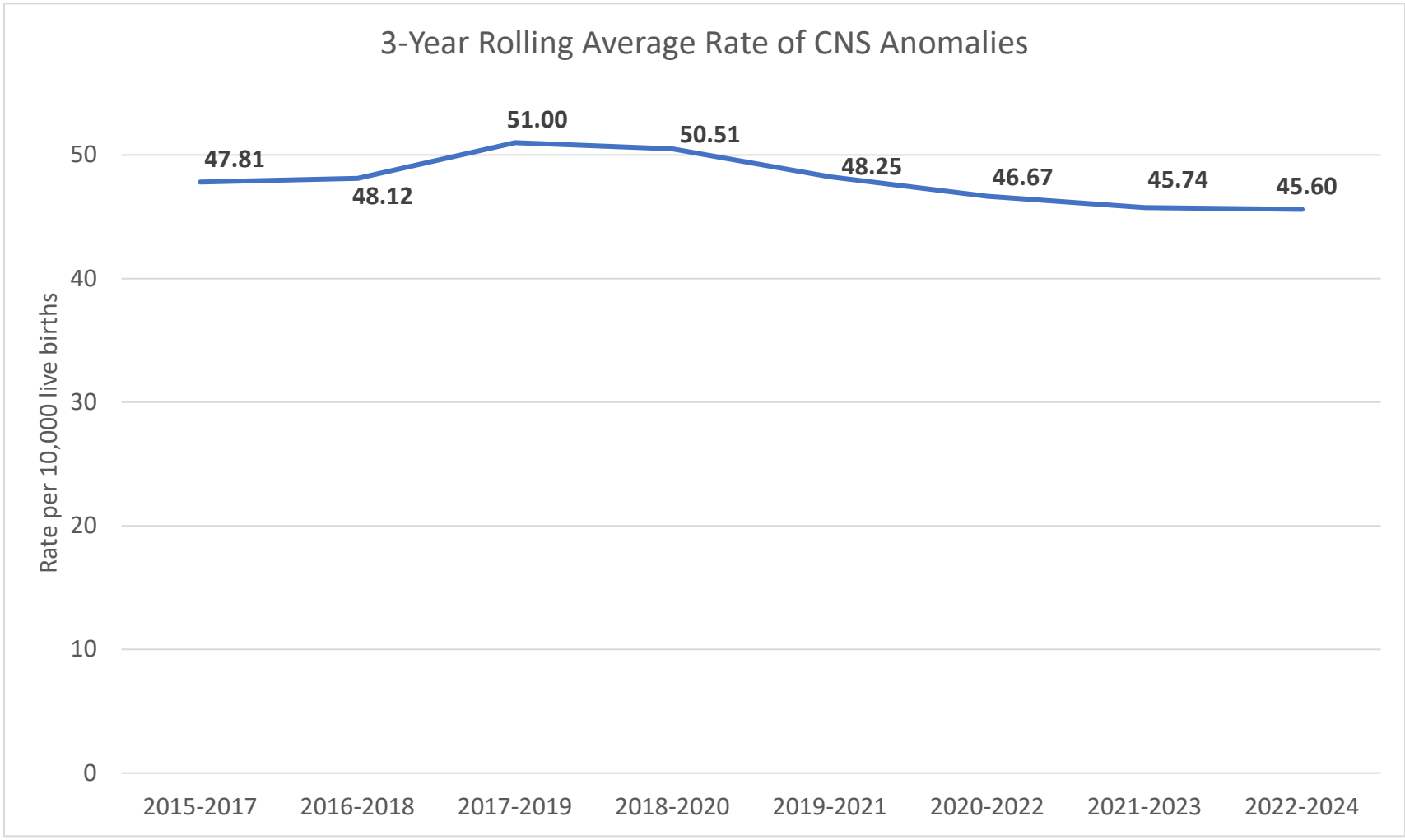

Figure 6. 3-Year Rolling Average NTD Rates

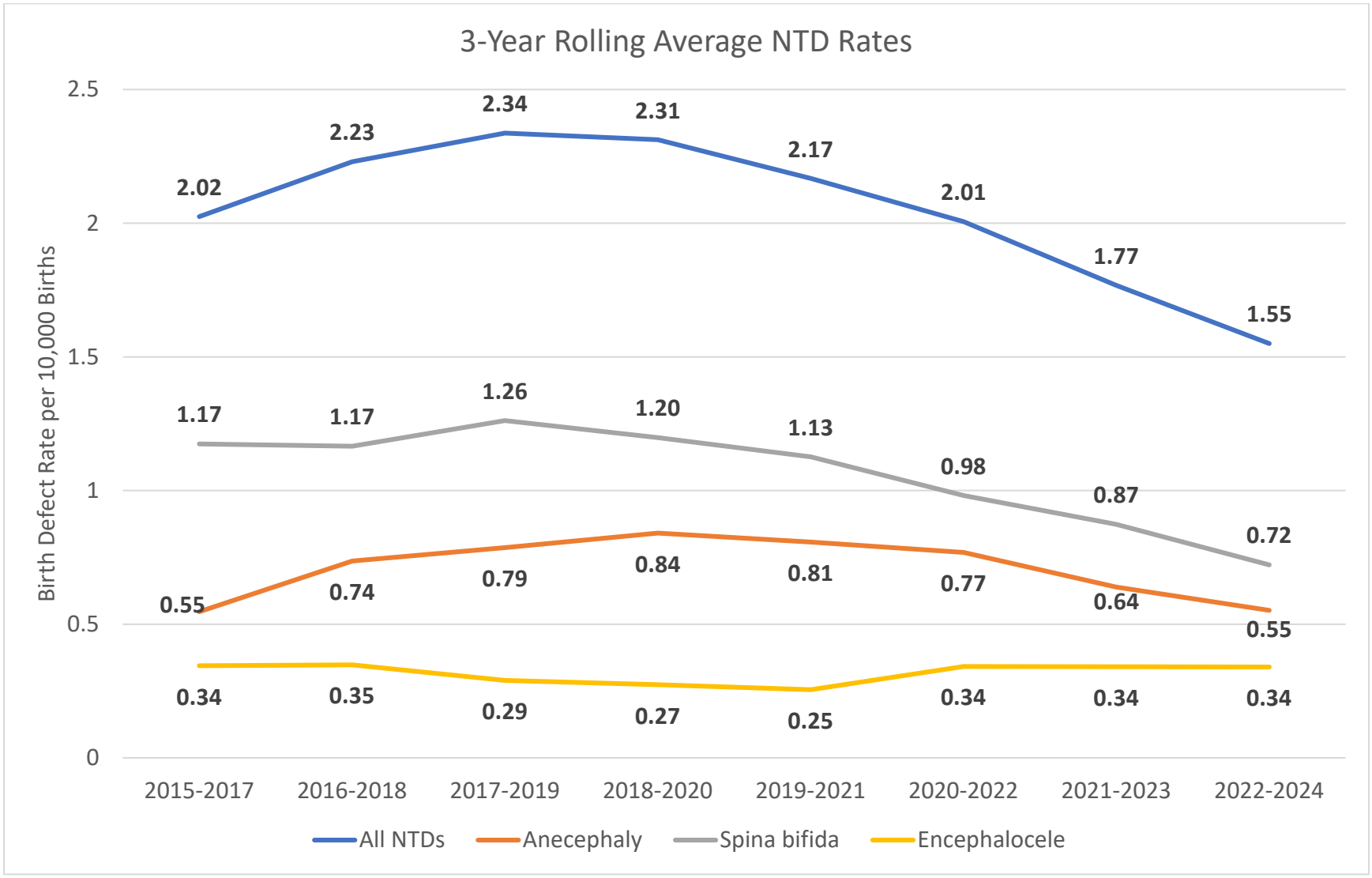

Figure 7. 3-Year Rolling Average Rate of Eye Anomalies

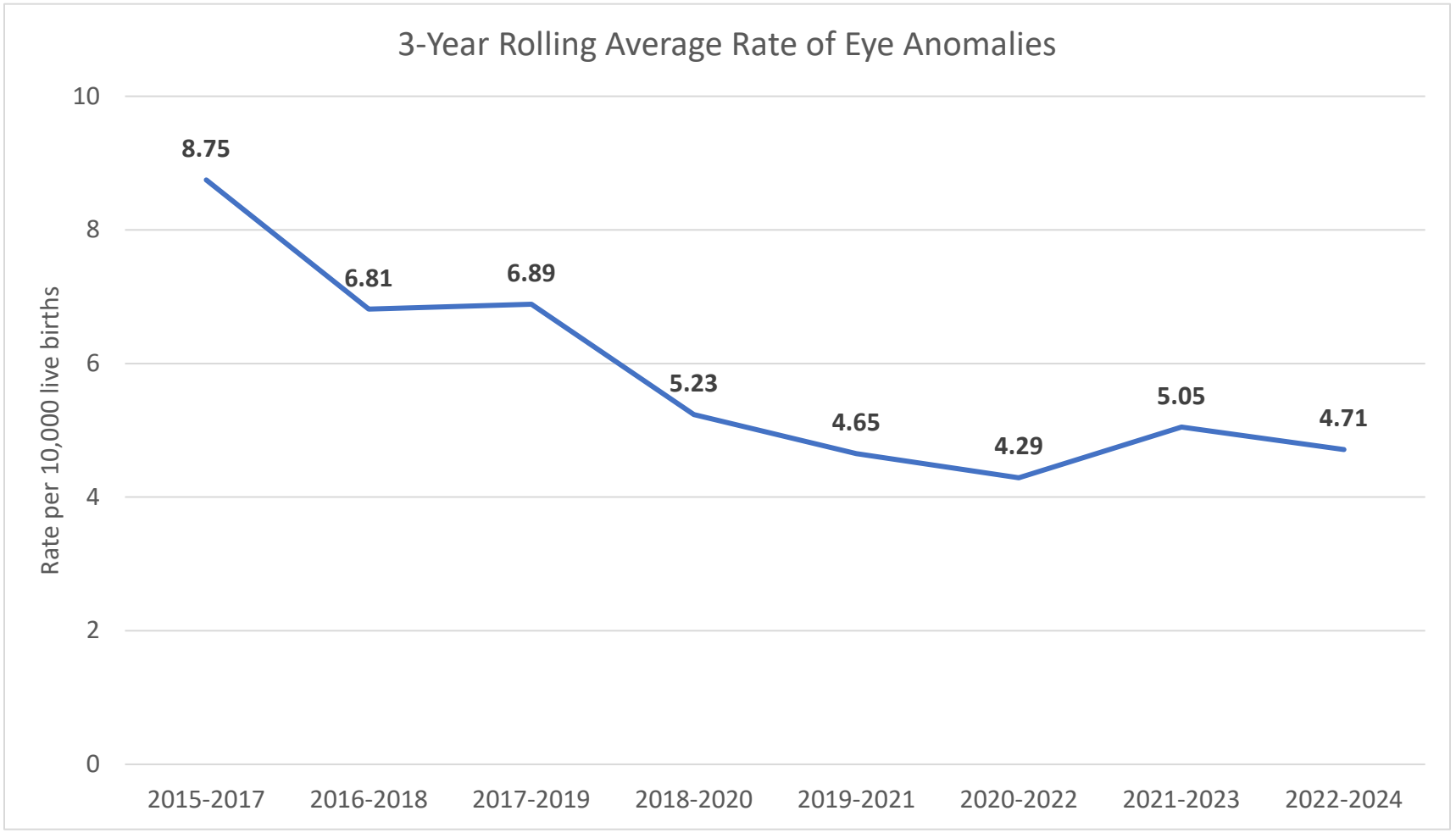

Figure 8. 3-Year Rolling Average Rate of Ear, Face, and Neck Anomalies

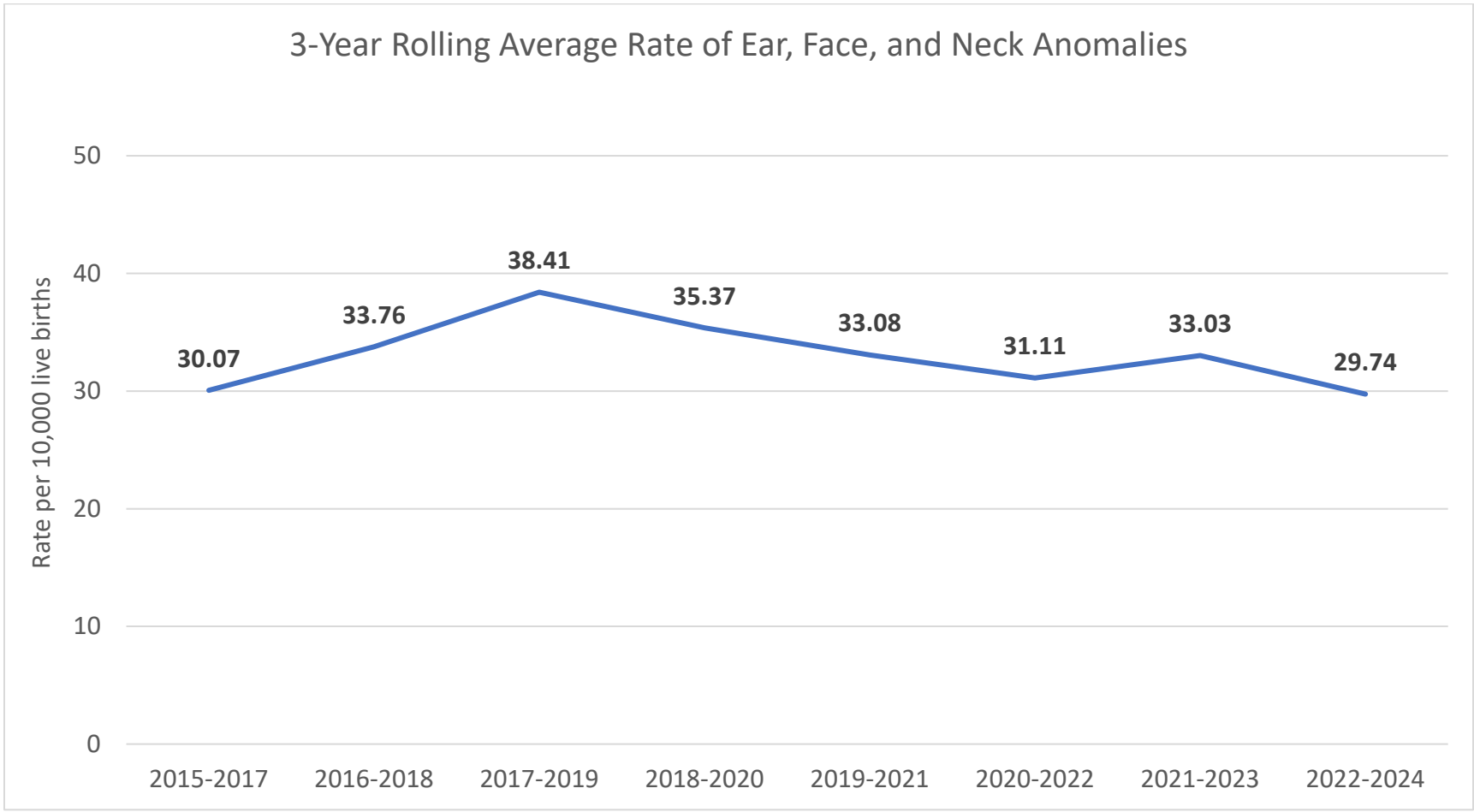

Figure 9. 3-Year Rolling Average of Cardiovascular Anomalies

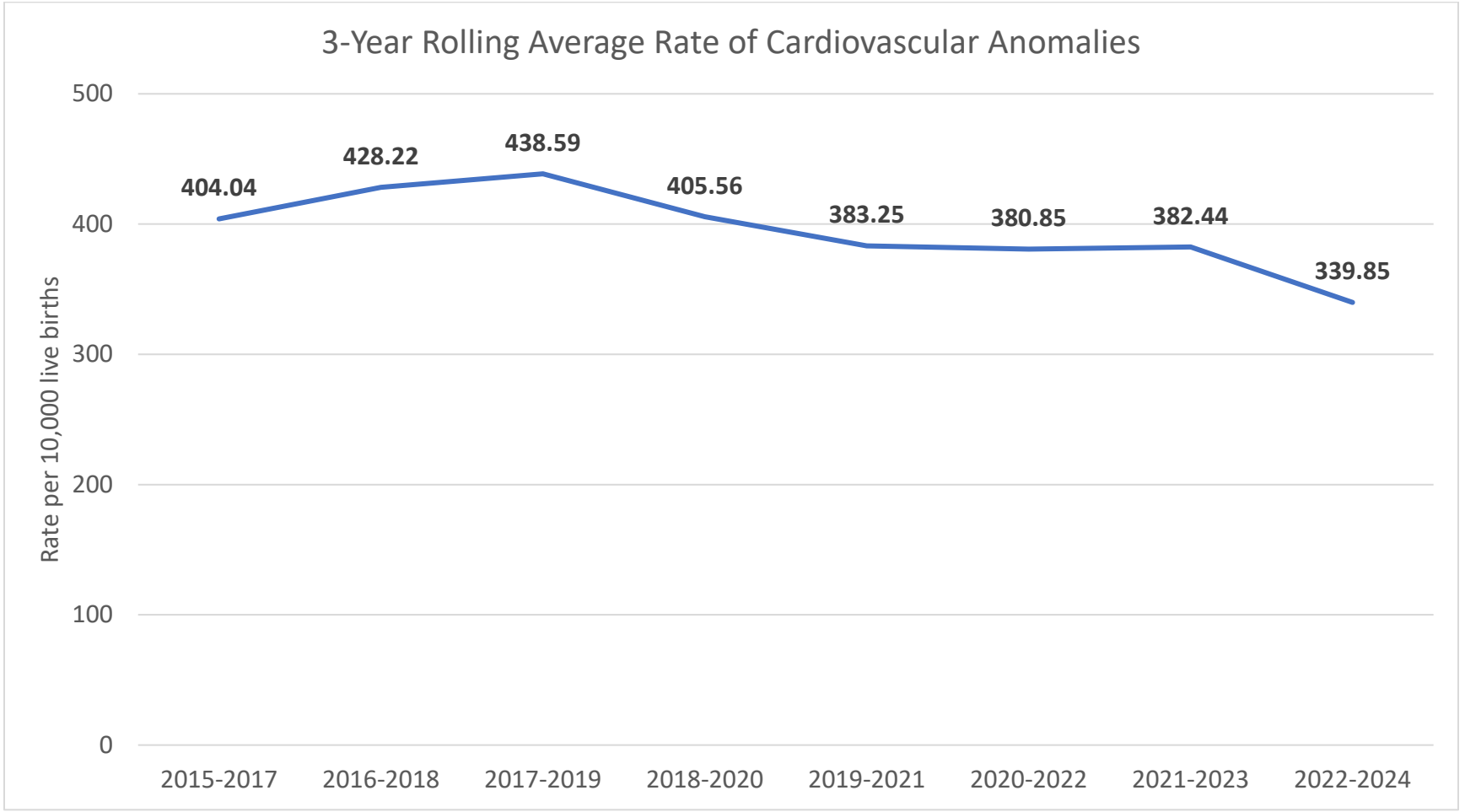

Figure 10. Rate of Primary and Secondary Lesions

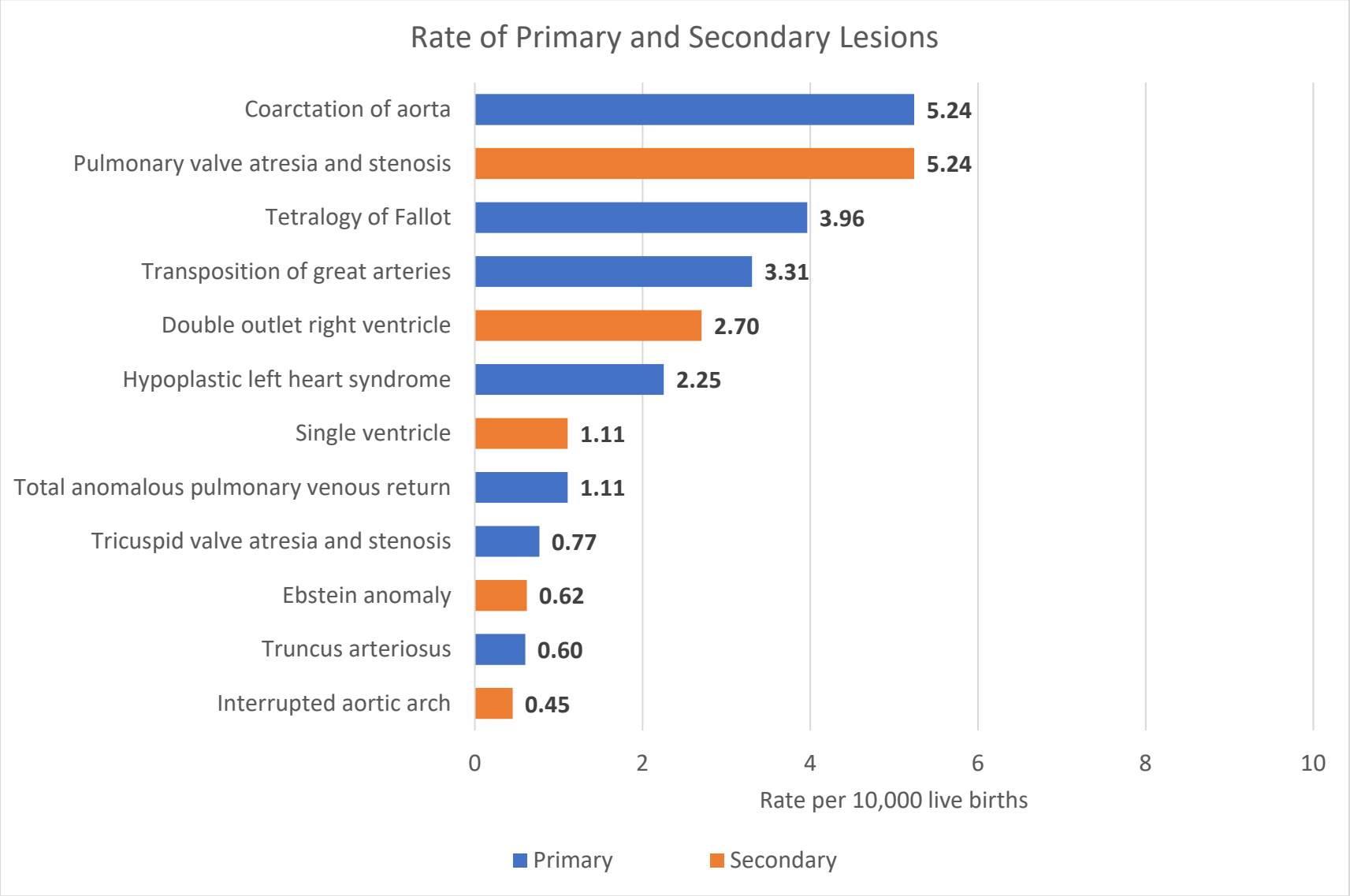

Figure 11. 3-Year Rolling Average Rate of Orofacial and Respiratory Anomalies

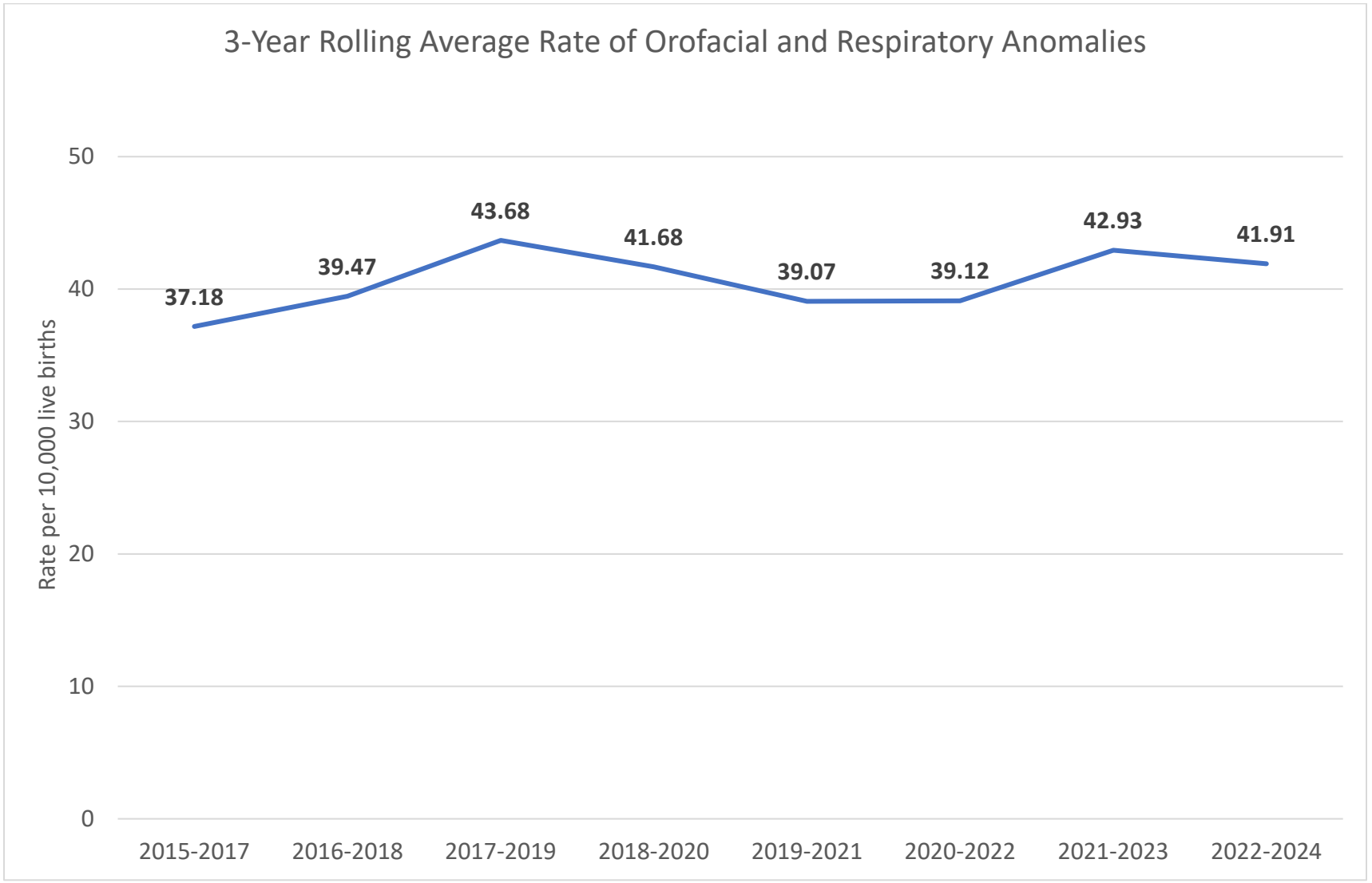

Figure 12. 3-Year Rolling Average Rate of Gastrointestinal Anomalies

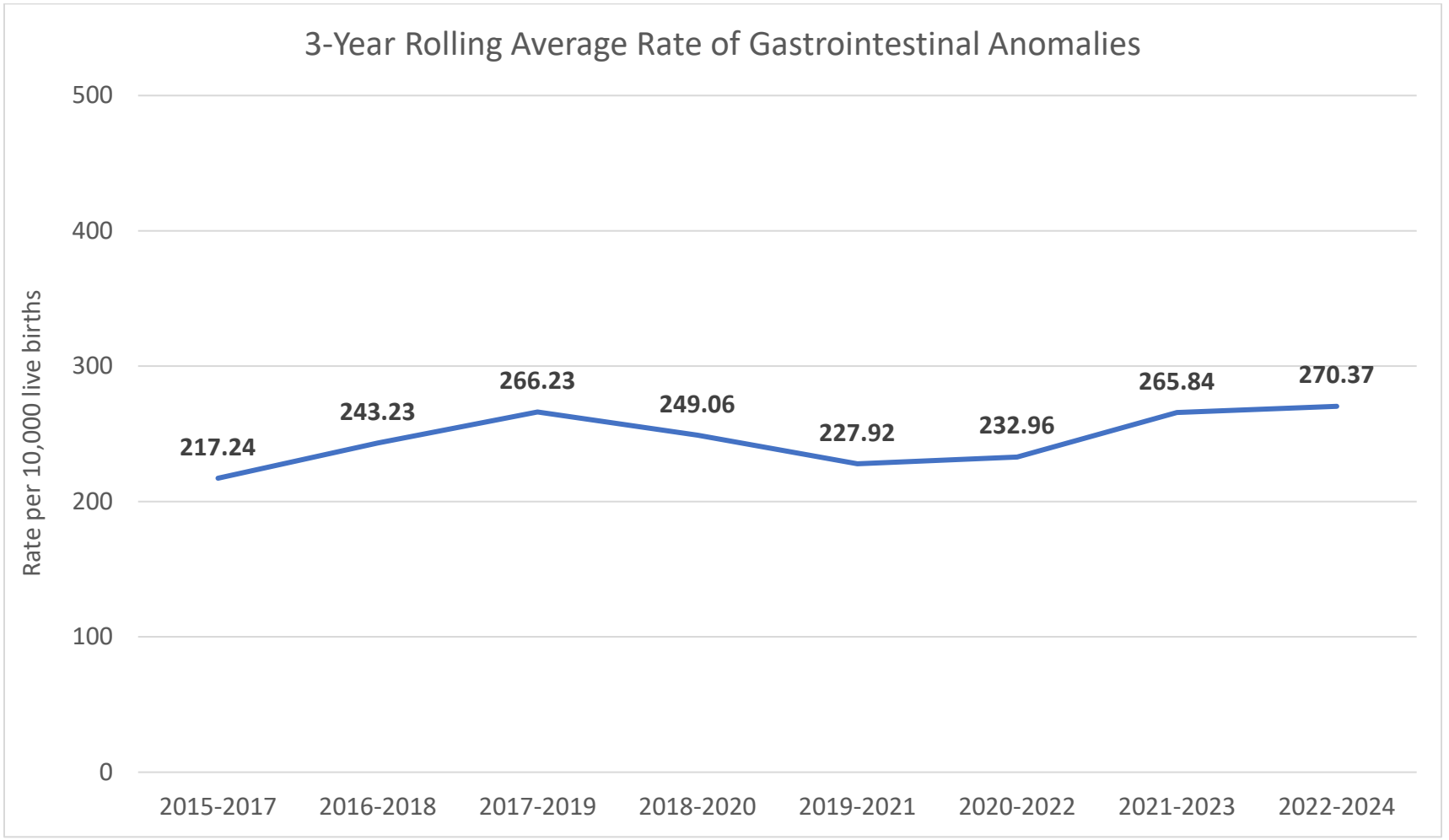

Figure 13. 3-Year Rolling Average Rate of Genitourinary Anomalies

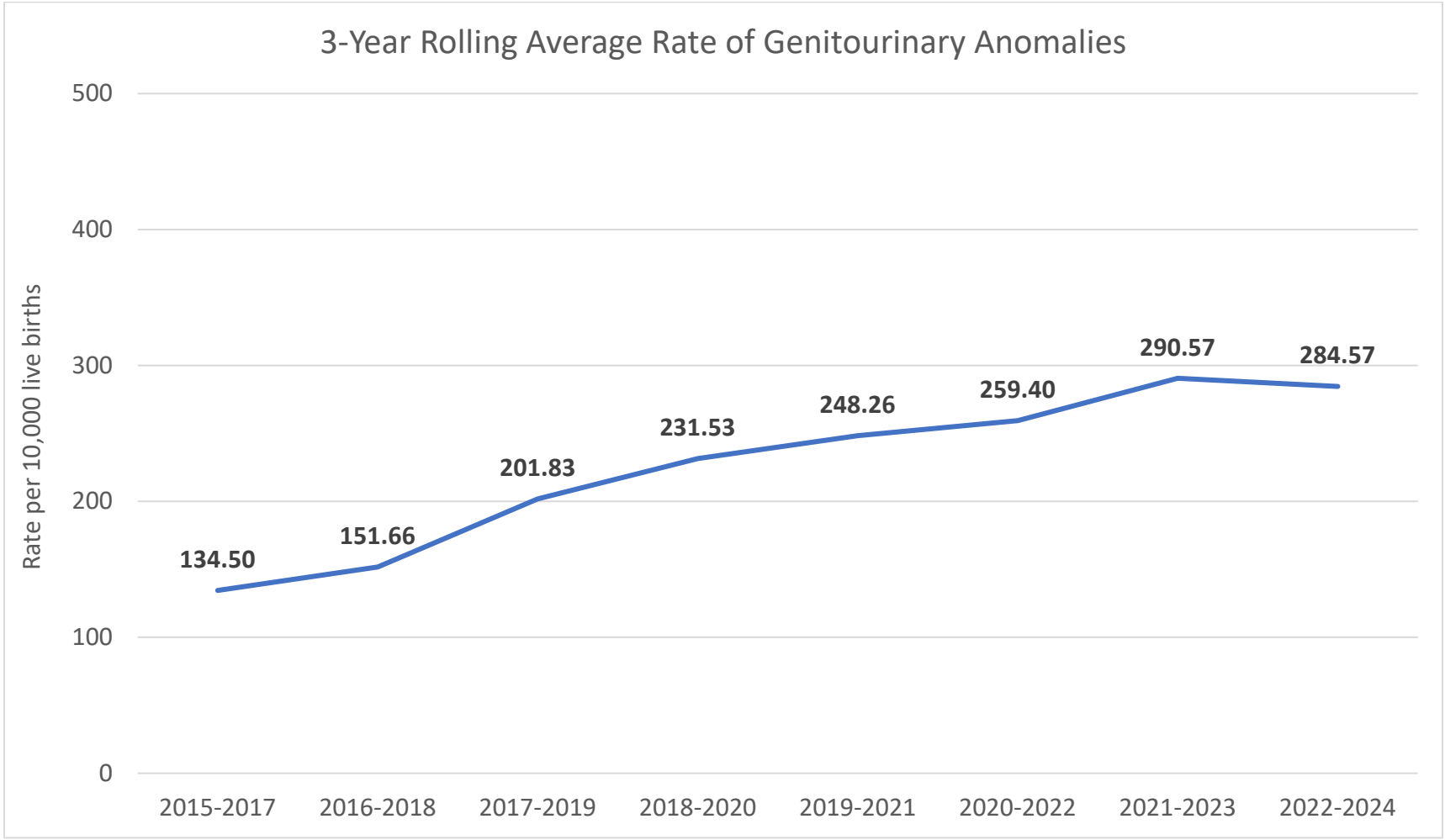

Figure 14. 3-Year Rolling Average Rate of Musculoskeletal and Limb Anomalies

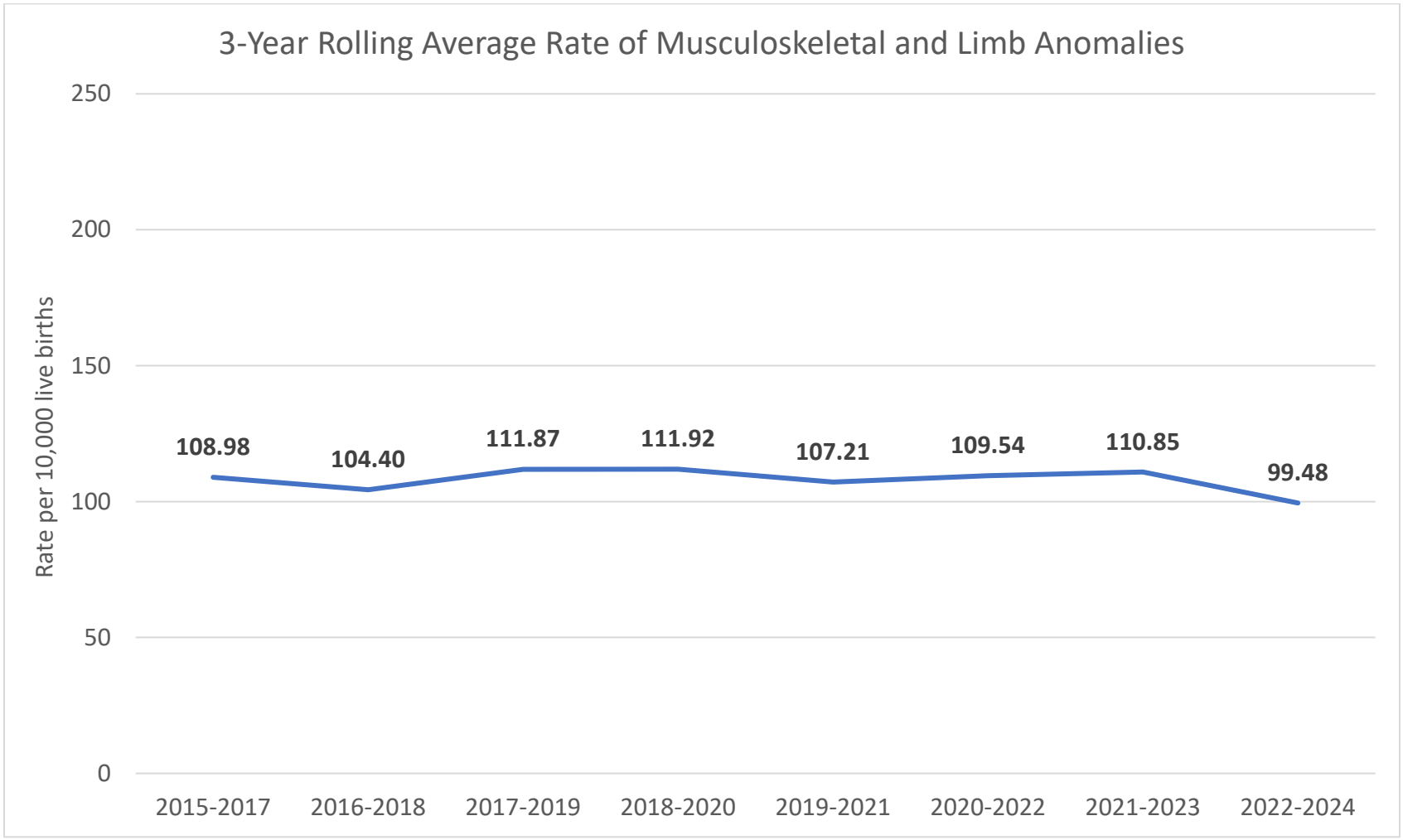

Figure 15. 3-Year Rolling Average Rate of Chromosomal Anomalies

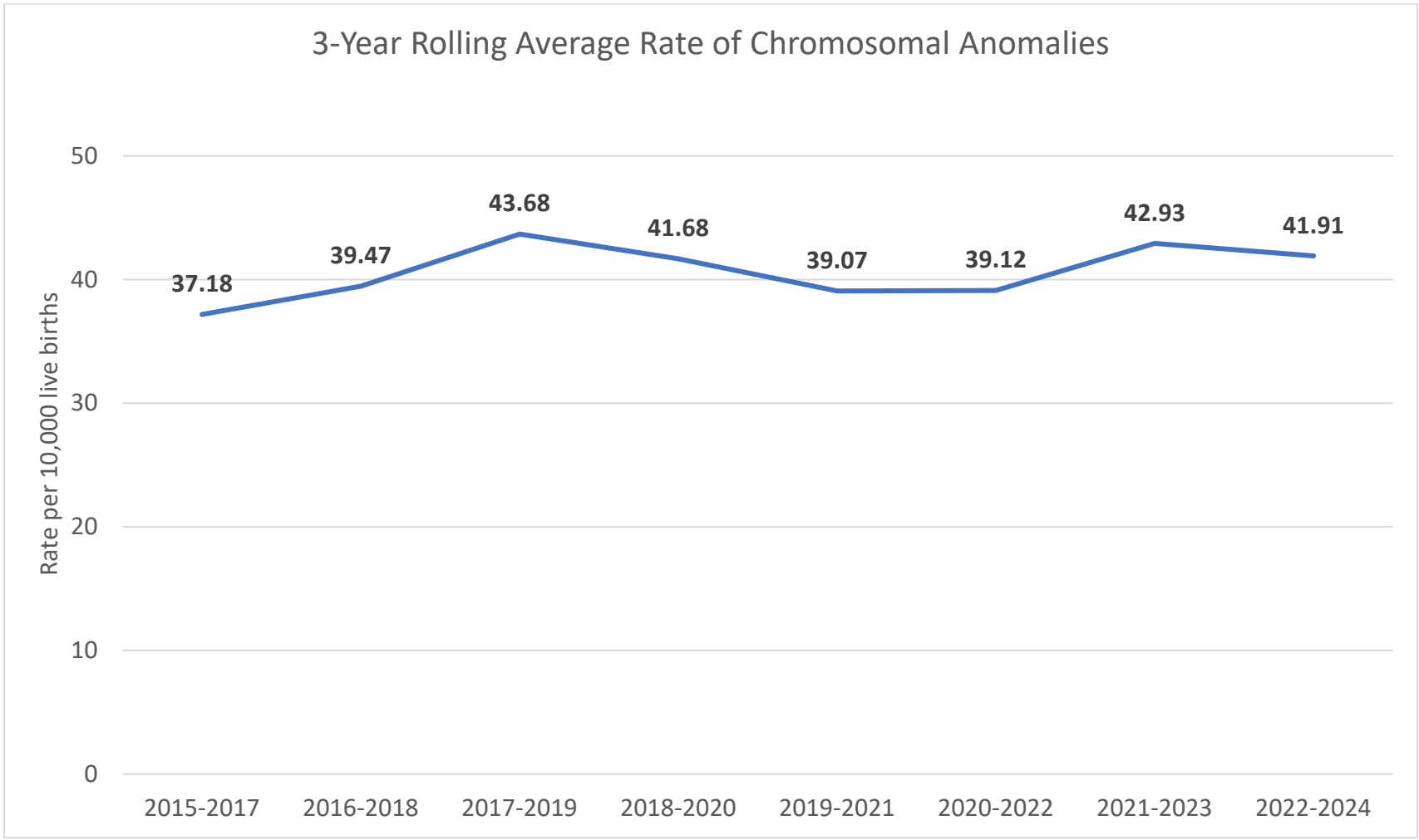

Figure 16. Gastroschisis by Maternal Age

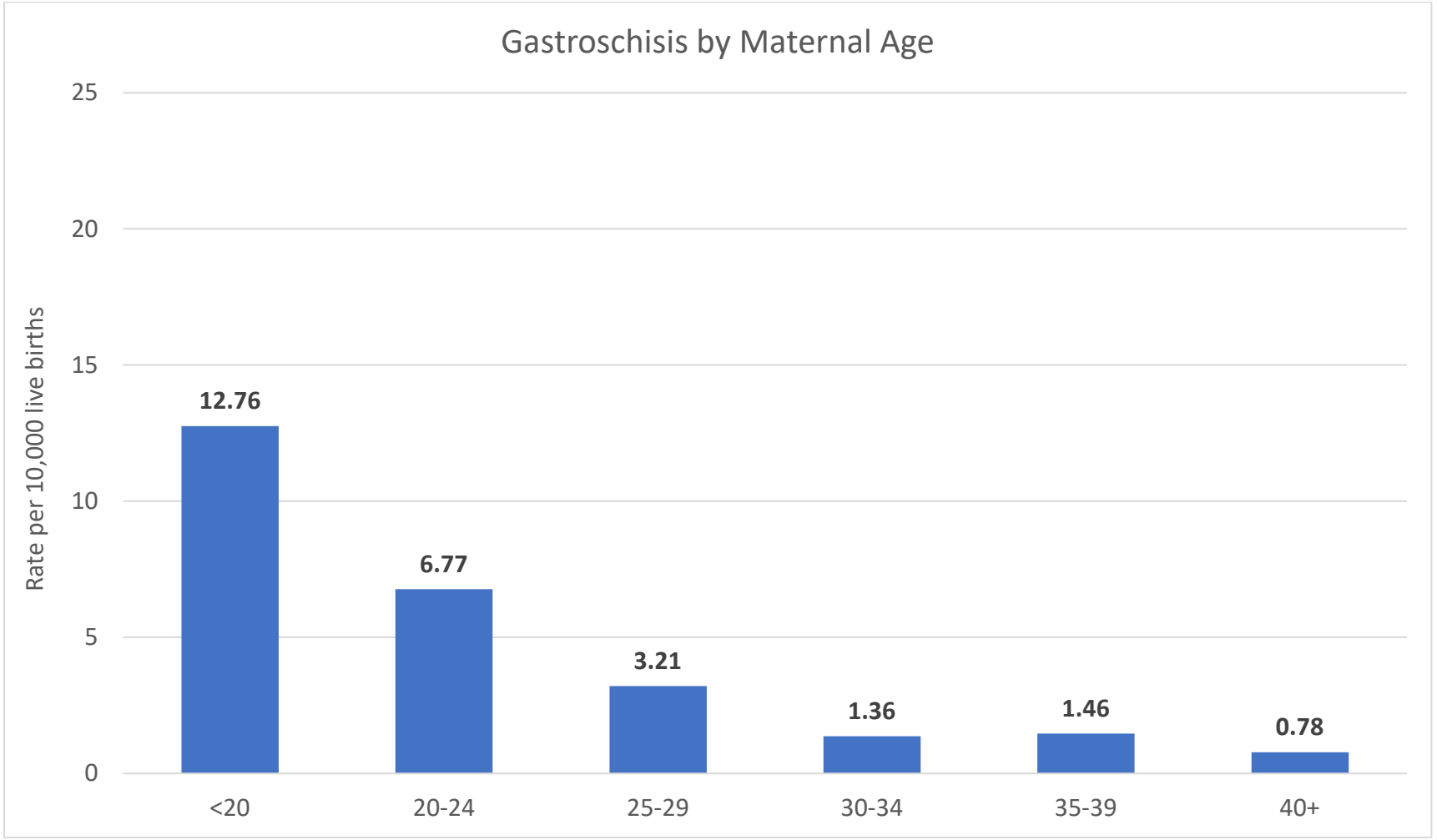

Figure 17. Trisomy 21 by Maternal Age

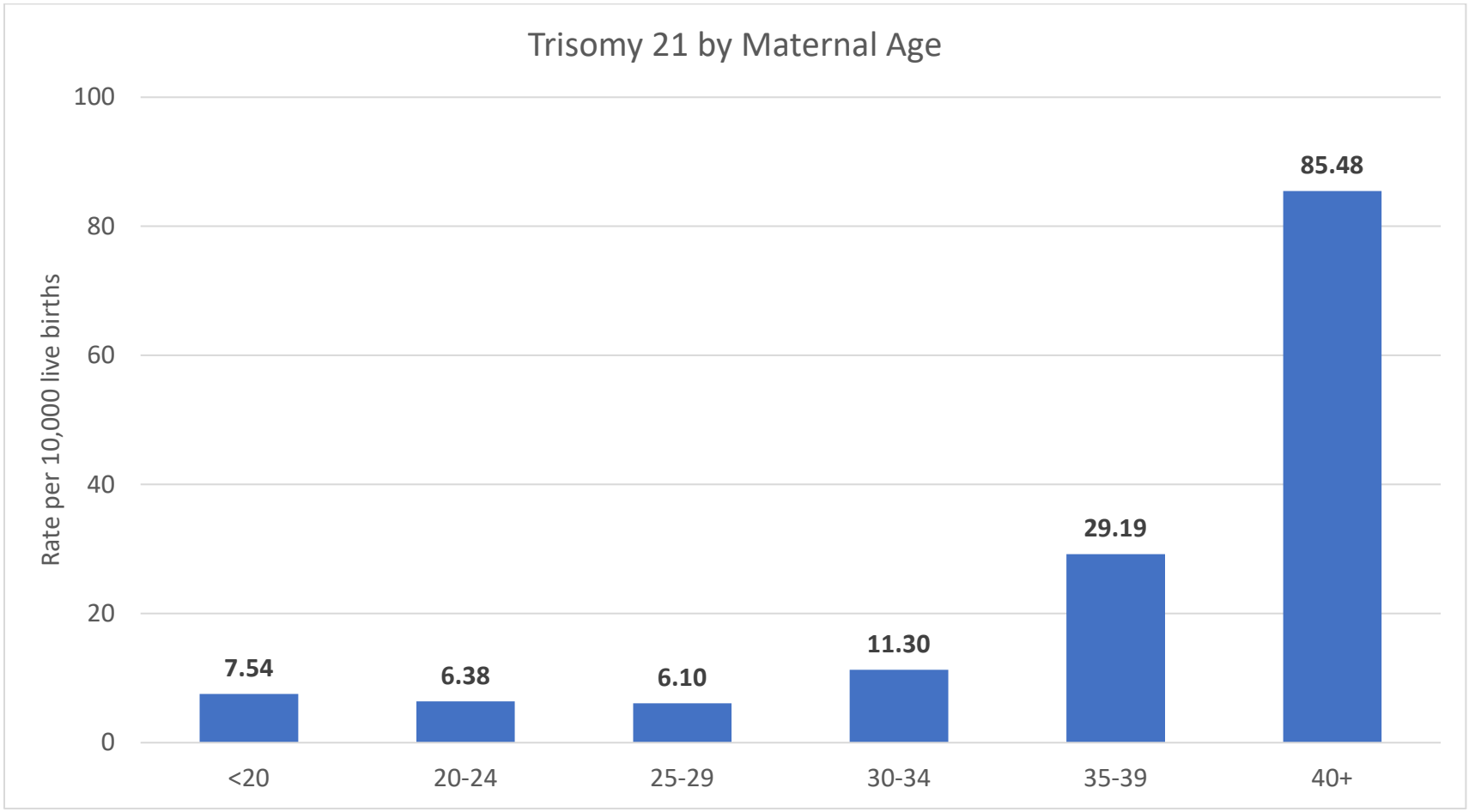

Figure 18. Frequency of Diabetes and Hypertension

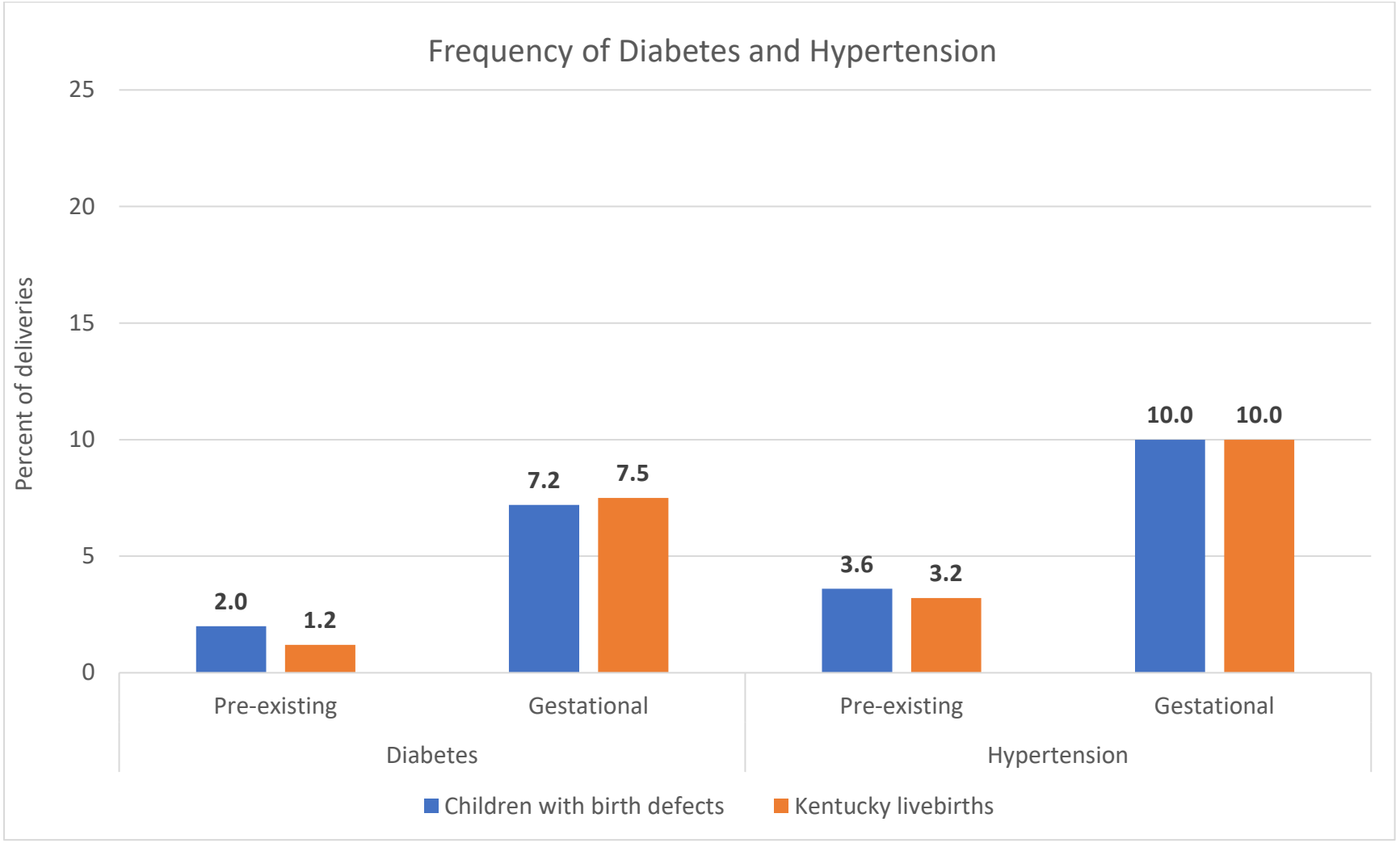

Figure 19. Increased Odds of Specific Conditions Associated with Pre-existing Maternal Diabetes

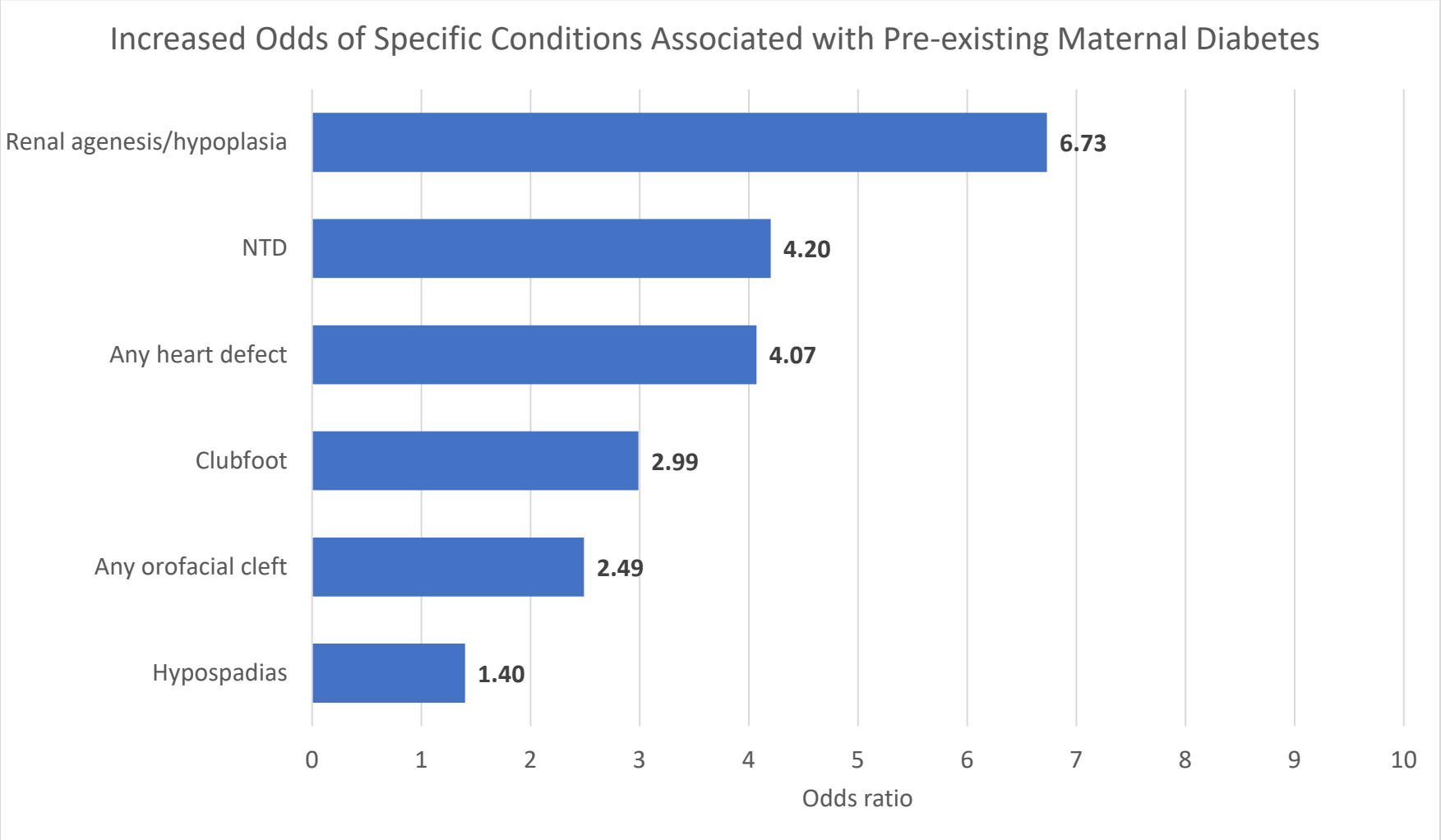

Figure 20. Increased Odds of Specific Conditions Associated with Pre-existing Maternal Hypertension

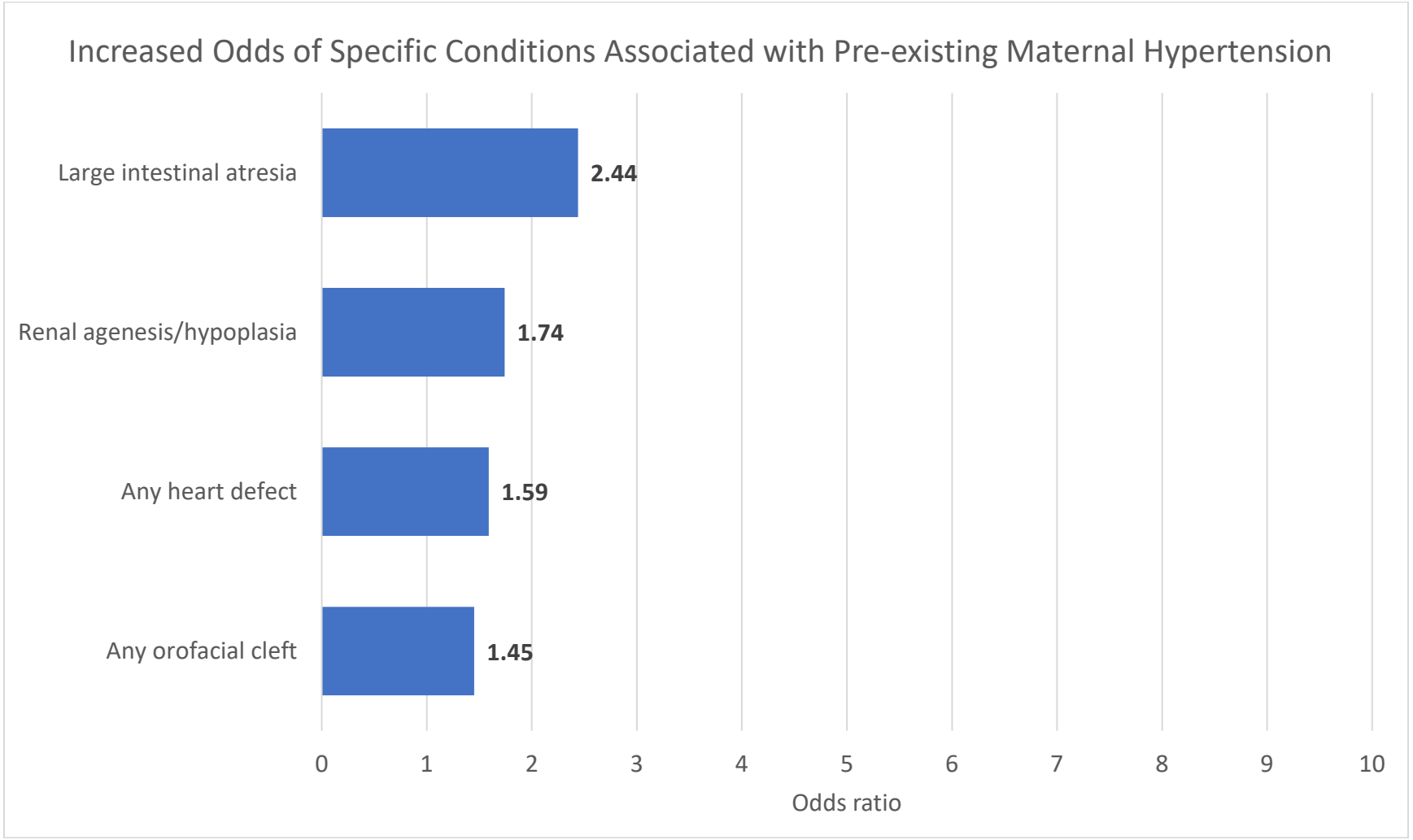

Figure 21. Gastroschisis by Pre-pregnancy BMI

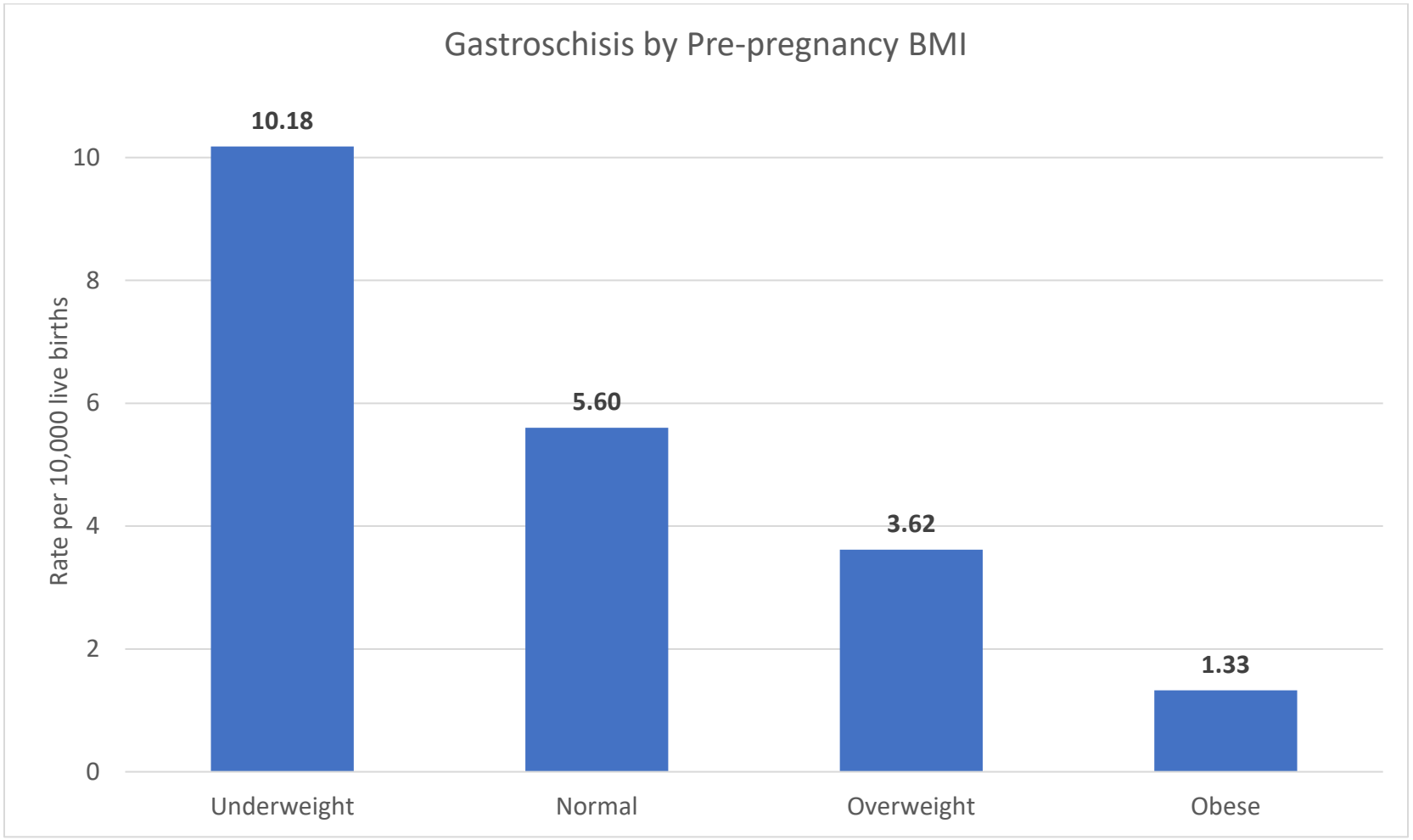

Figure 22. Increased Odds of Specific Conditions Associated with Maternal Smoking

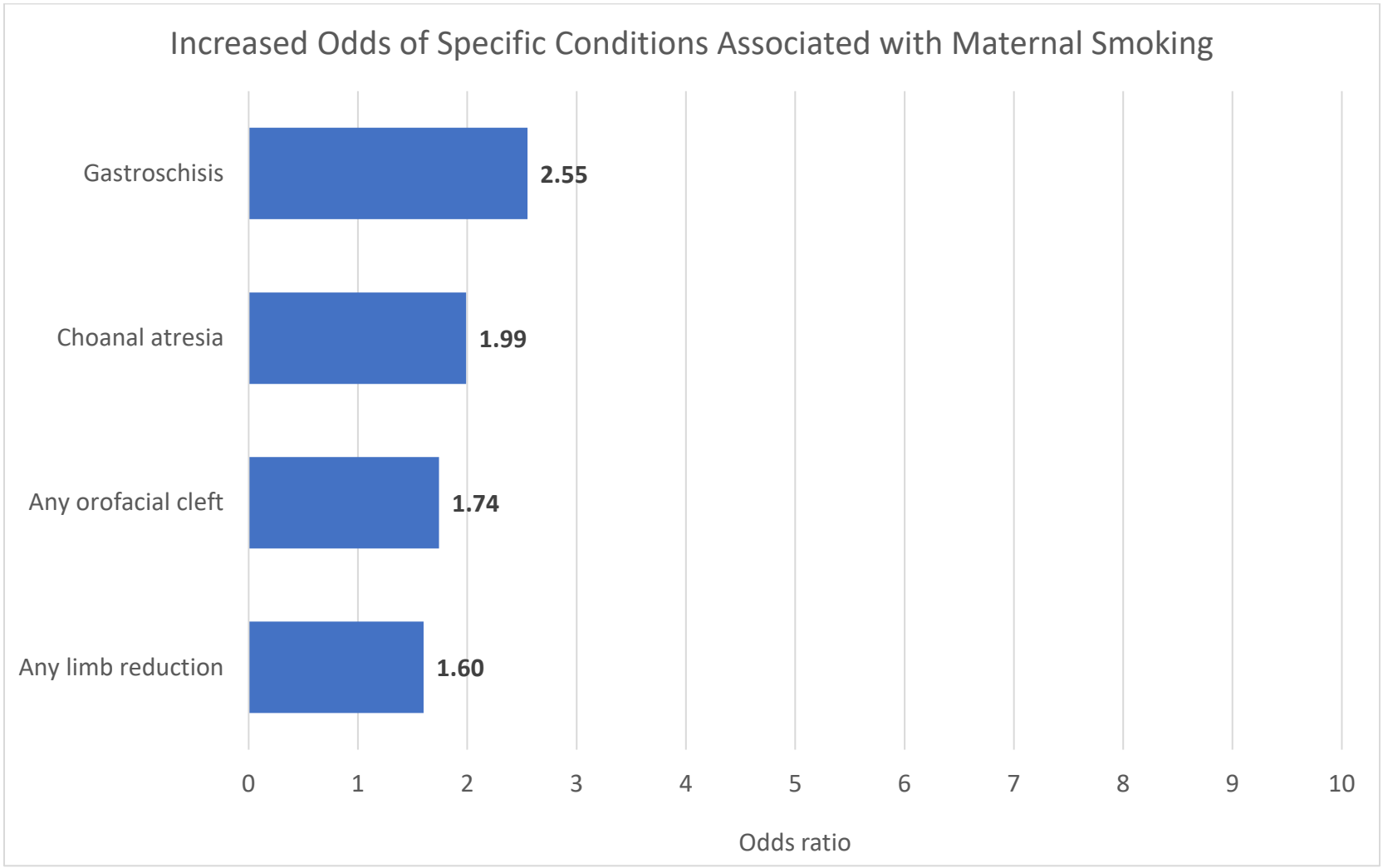

Figure 23. Rate of Cardiovascular Defects by Maternal Smoking

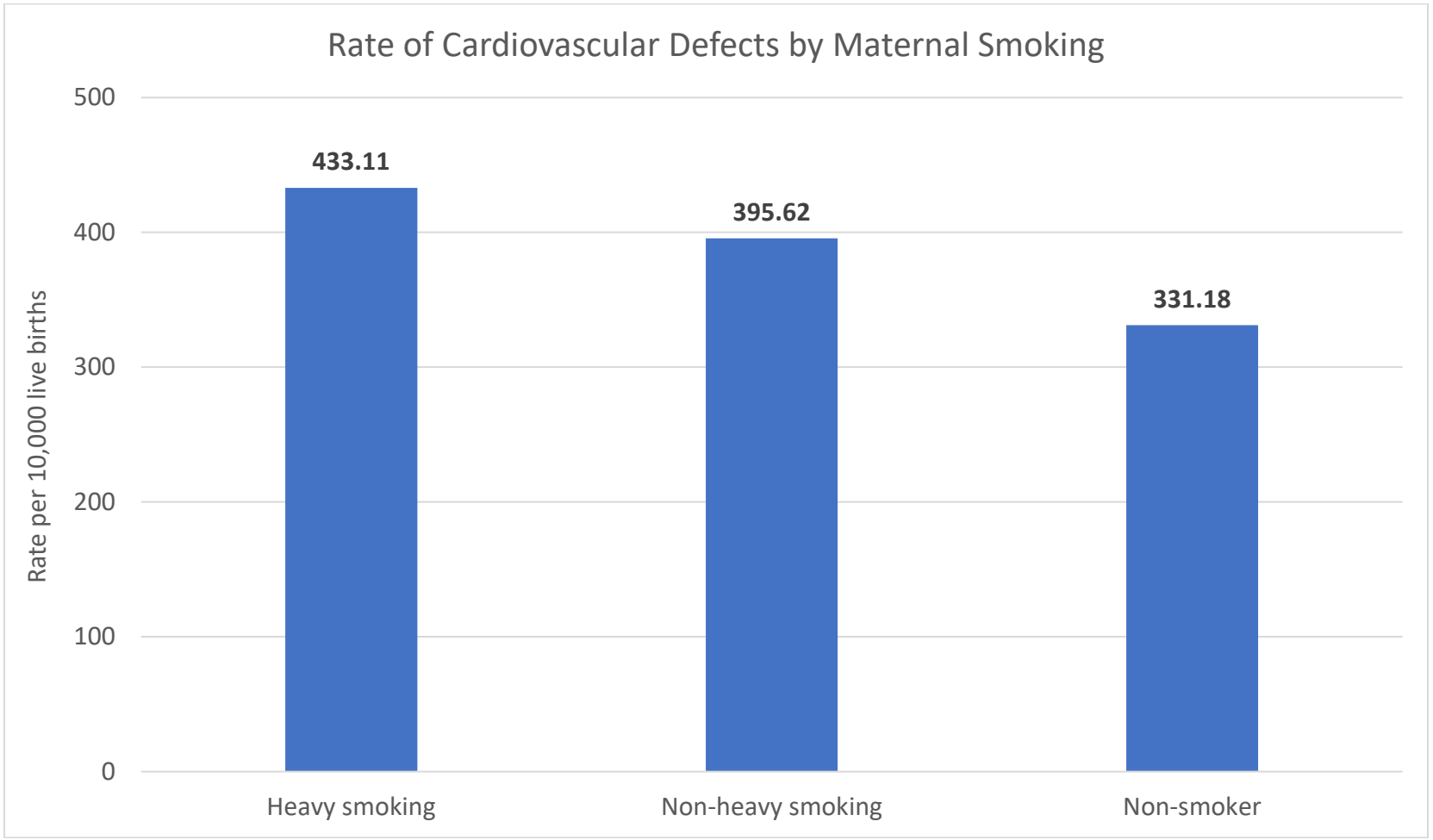

Figure 24. Frequency of Preterm Birth Categories

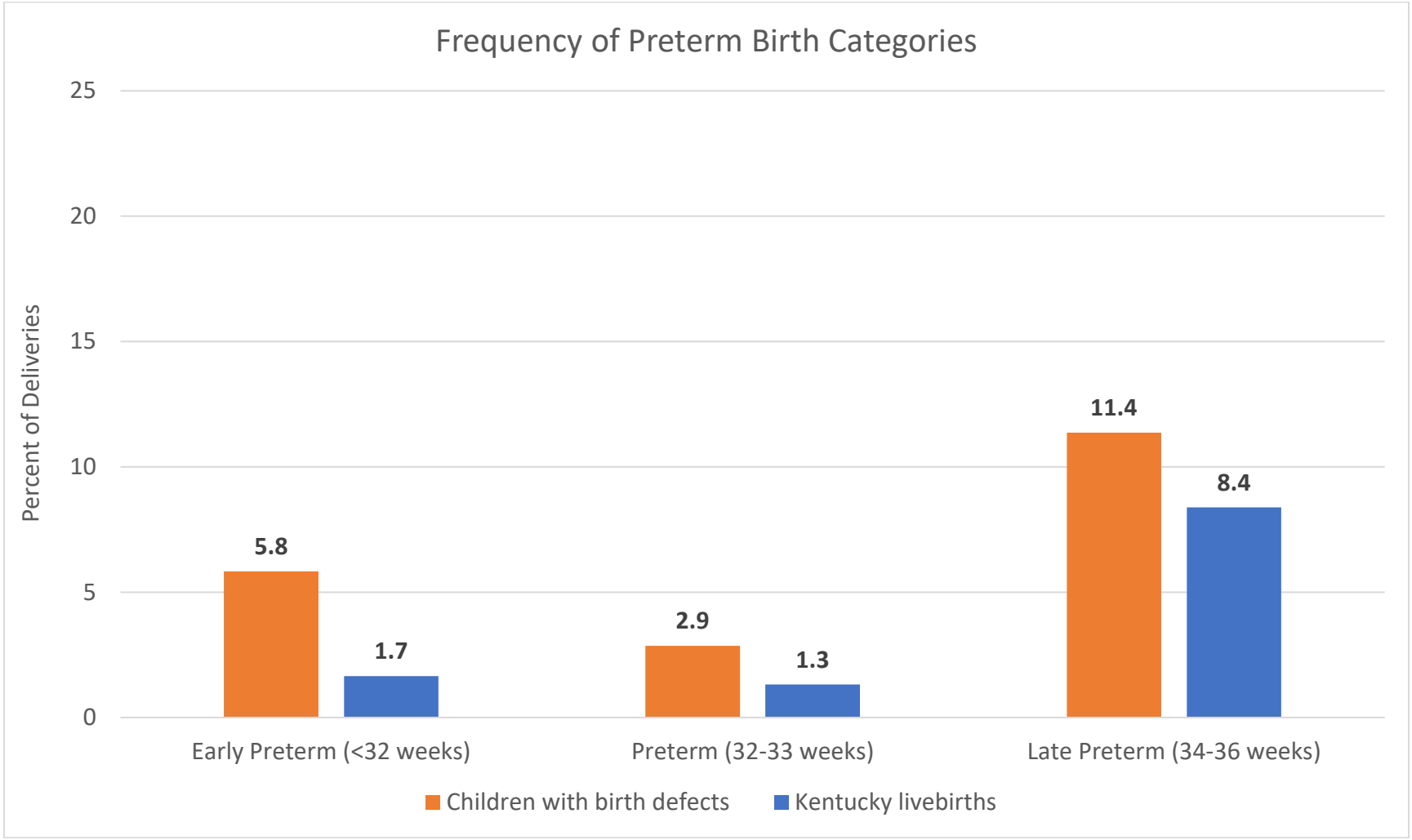

Figure 25. Rate of Cardiovascular Defects by Gestational Age

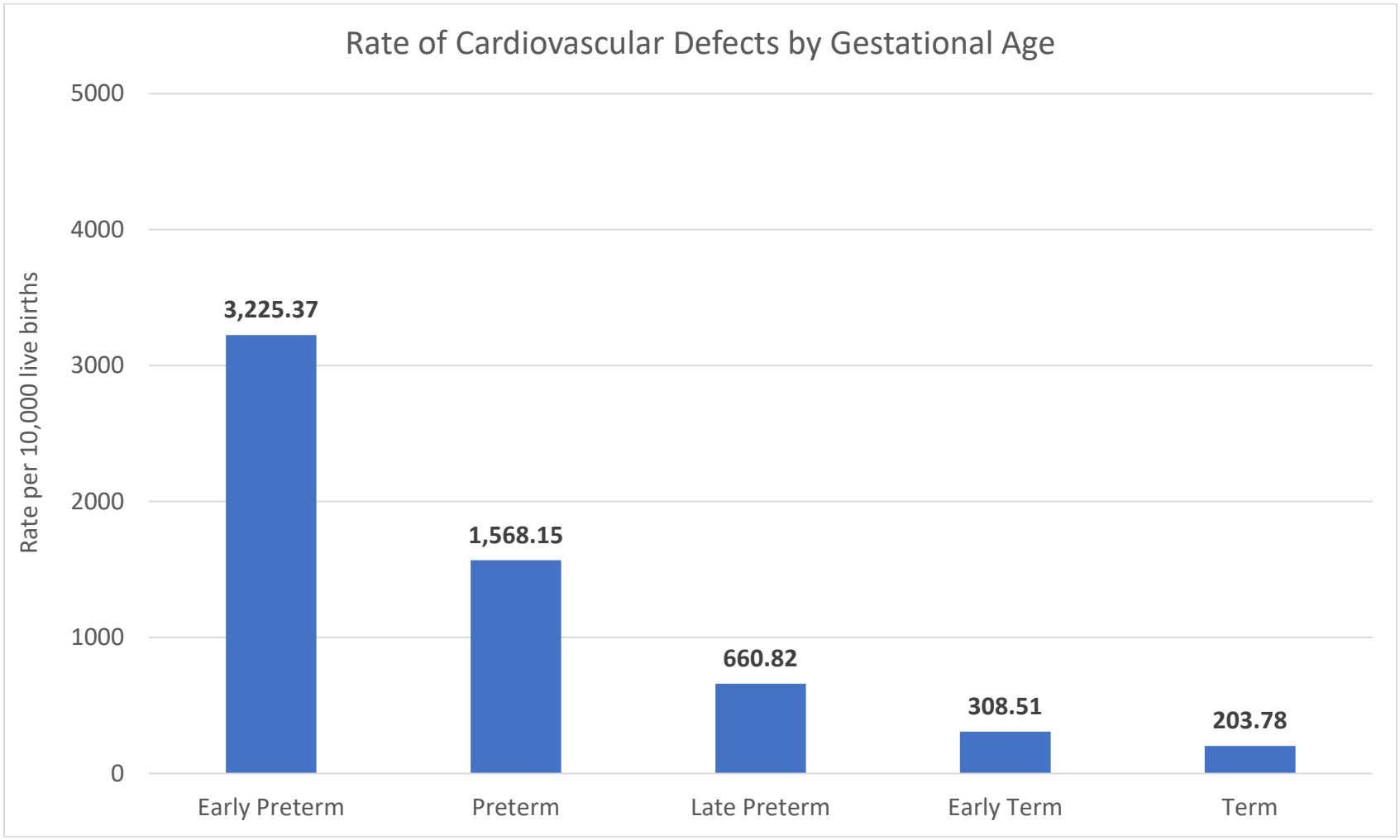

Figure 26. Most Common Types of Birth Defects Documented as Causing Death or Stillbirth

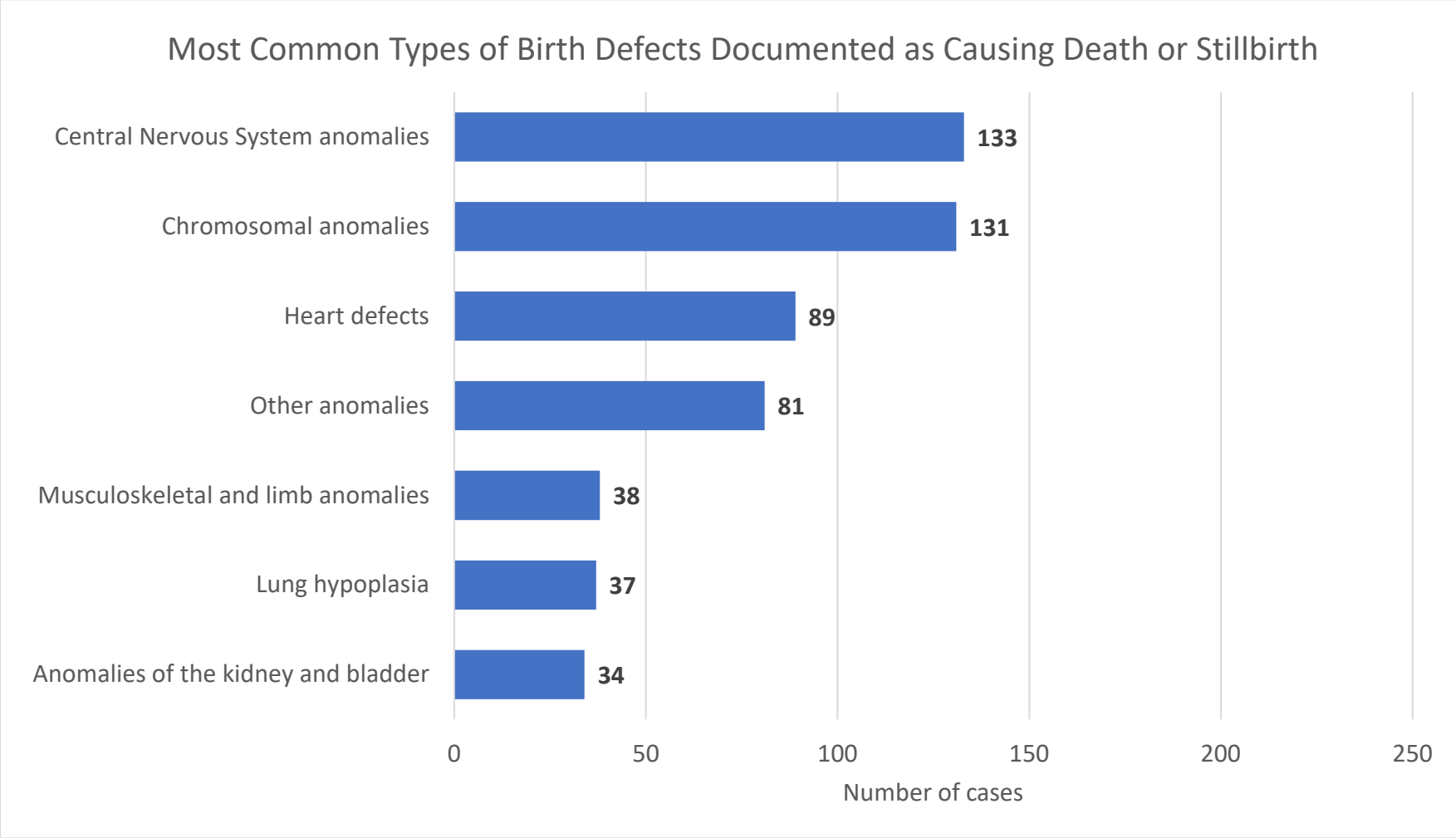

Figure 27. Birth Defects with the Highest Fatality Rates