



**Kentucky Department for Public Health  
Division of Maternal and Child Health**

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August 2016

# **Ten Years of Prevention**

## **The Kentucky Birth Surveillance Registry**

### **2005-2014**

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**Kentucky Department for Public Health  
Division of Maternal and Child Health**

**August 2016**



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### Note from the Director

Birth defects are common, costly, and most importantly may have a significant impact on families. As Kentucky's Maternal and Child Health Division Director, I recognize the importance of the birth defects surveillance program in fulfilling the Division's mission to "provide leadership, in partnership with key stakeholders, to improve the physical, socio-emotional health, safety and well-being of all Kentucky women, infants, children, adolescents and their families." The data collected by KBSR is used to enhance prevention activities, make referrals for needed services such as early intervention, and evaluate programs. This 10-Year Report is a culmination of KBSR data that is being released to our partners and the public with the goal of providing education about birth defects in Kentucky and emphasizing prevention strategies. It is my hope that through data sharing we can work together to better understand birth defects, ensure that families affected by birth defects have the services they need, and complete activities to prevent birth defects.

Sincerely,



Ruth Ann Shepherd, MD, FAAP  
Director, Division of Maternal and Child Health

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This report may be viewed at the following web address: <http://chfs.ky.gov/dph/mch/ecd/kbsr.htm>

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## **Birth Defects**

### **What are Birth Defects?**

Birth defects (or congenital anomalies) are health conditions that alter the structure of one or more body parts and occur during development in pregnancy.<sup>1</sup> Birth defects affect how the body looks, works, or both. Some problems are very minor and may require no interventions or can be corrected with surgery or therapy, while other birth defects can cause lifelong physical and/or mental disabilities. A birth defect may not affect the expected lifespan of the child; however, some birth defects can impact lifespan depending on the severity.

### **How Common are Birth Defects?**

About 120,000 babies are born with a major birth defect in the United States (U.S.) every year, which is the equivalent of 1 in every 33 births.<sup>1</sup> In the Commonwealth of Kentucky, birth defects are even more common. Each year, about 4,900 babies (about 1 in every 12 births) are born with a birth defect, including major and minor birth defects, and those that are transient (naturally resolve on their own).

Birth defects are a common cause of mortality among infants (babies under 1 year old). In Kentucky, about 20% of all infant deaths are caused by birth defects, which is the equivalent of about 75 cases per year.<sup>2</sup> Birth defects are also implicated in about 45 stillbirths per year in Kentucky (about 13% of all reported stillbirths).

### **Prevention**

Not all birth defects can be prevented, but there are ways that a woman can increase her chance of having a healthy baby. She can quit smoking, not drink alcohol, and avoid drugs and chemicals. She can discuss any chronic health conditions (such as diabetes or high blood pressure) and medication use (including prescription, over-the-counter, and supplements) with her doctor.<sup>1</sup>

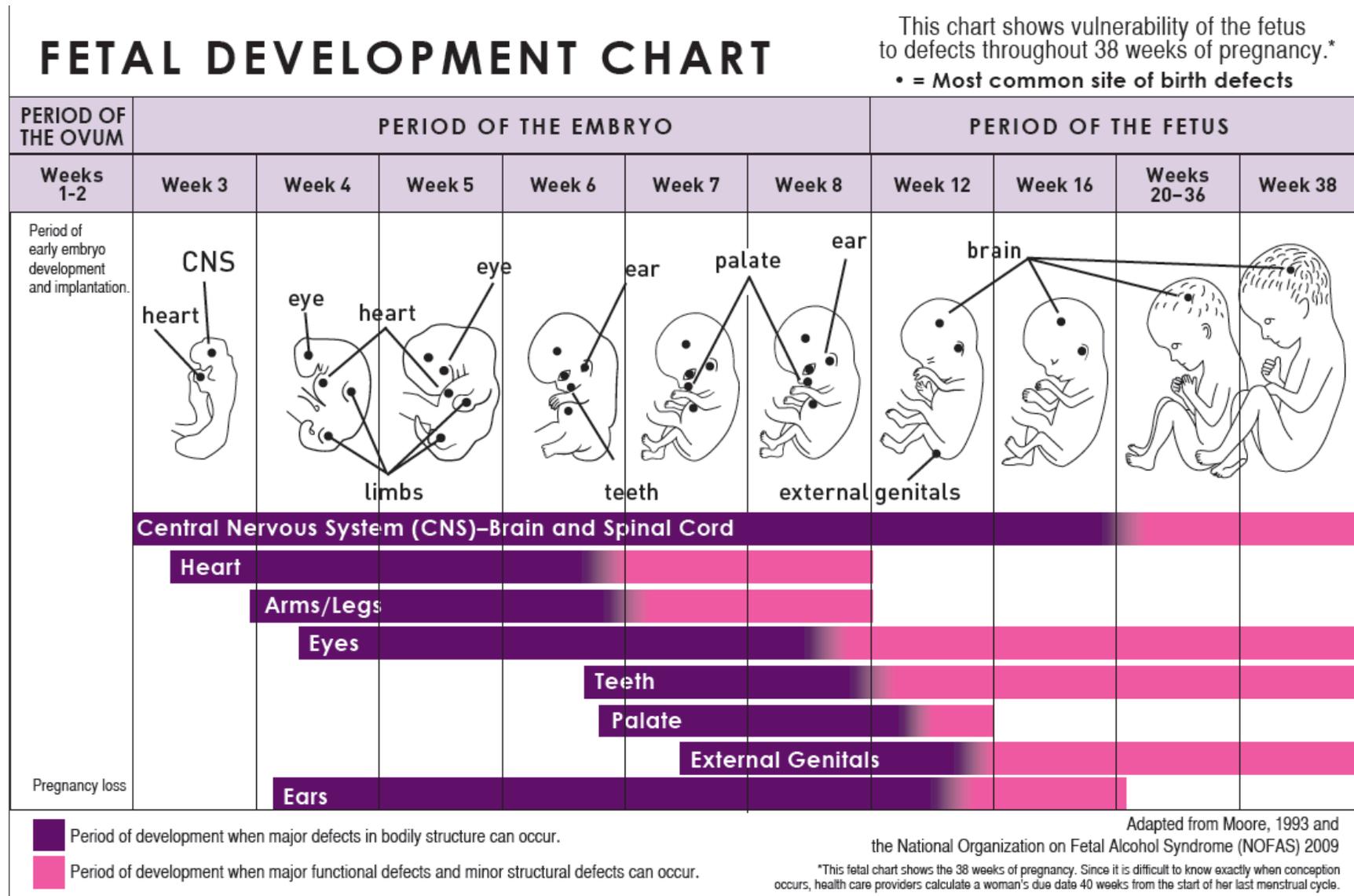
The causes of most birth defects are unknown; there is a complex mix of risk factors, including genetics, behaviors, and environmental exposures.<sup>1</sup> Having a risk factor does not guarantee that a pregnancy will be affected by a birth defect, and even women with no known risk factors can have a baby with a birth defect. More prevention information is found on the following page.

### **Identification and Living with a Birth Defect**

Birth defects may be identified at different times, including before birth, at birth, or after birth. However, most are identified within infancy.<sup>1</sup> Some birth defects are visible, but others must be diagnosed with specialized tests or equipment. Accurate identification of birth defects is critical for planning to meet the child's medical and developmental services.

Babies who have a birth defect often need special care to survive and thrive, and their parents might find themselves feeling lost or in need of support. Fortunately, there are many resources available to Kentucky families, some of which are presented in this report. For more information about early intervention and other services, please see the Partners section on page 45.

Figure 1: Fetal Development Chart Showing Vulnerability of Fetus to Defects Related to Substance Exposure During Pregnancy



## Centers for Disease Control and Prevention's Top 10 Tips for Preventing Birth Defects

Many birth defects occur during the first few weeks of pregnancy.<sup>1</sup> About 49% of pregnancies in the U.S. are unintended.<sup>3</sup> Because a woman might not know she is pregnant during the crucial first weeks of pregnancy, there can be difficulties associated with efforts to reduce the risks of birth defects.

The best way to prevent birth defects is for parents to stay healthy before and during a pregnancy, including<sup>4</sup>:

1. Take a multivitamin with at least 400 micrograms of folic acid daily beginning at least 1 month before a pregnancy and continuing throughout the duration of the pregnancy.
2. Visit a health care provider regularly. Early and consistent prenatal care is one way that a woman can monitor her health during pregnancy.
3. Abstain from drinking alcohol. There is no known safe time, amount, or type of alcohol to drink during pregnancy.
4. Quit smoking before or during a pregnancy and avoid second-hand smoke. Tobacco smoke makes premature birth (<37 weeks), certain birth defects, and infant death more likely to occur.
5. Don't use street drugs during pregnancy. Illegal drugs increase the risk of preterm births and low birth weight infants and can cause heart and limb birth defects.
6. Prevent infections by washing hands, avoiding animals and animal excrement, and eating food that has been well-cooked or pasteurized.
7. Control diabetes during pregnancy. Diabetes can increase the risk of birth defects and other problems for a baby and mother.
8. Reach and maintain a healthy weight. Obesity has been shown to increase the risk of birth defects and other serious problems in pregnancy.
9. Talk to a health care professional about all medications, whether they are dietary or herbal supplements, prescription drugs, or over-the-counter medications. Some medications can cause serious birth defects. However, don't stop or change medications that are necessary without talking first with a health care provider.
10. Talk to a healthcare provider about vaccinations. Most vaccinations are safe during pregnancy, and some are even recommended during this time. Many infectious diseases are more severe for pregnant women, and others can cause birth defects, so vaccination is especially important.

Not all birth defects can be prevented, but simple changes can reduce the risk of having a baby with a birth defect.

# The Kentucky Birth Surveillance Registry

## What is the Kentucky Birth Surveillance Registry (KBSR)?

KBSR is a state-mandated surveillance system designed to provide information on the incidence, prevalence, trends, and possible causes of stillbirths, birth defects, and disabling conditions. KBSR operates under the authority of Kentucky Revised Statute (KRS) 211.651-670 (Appendix 1). Since 1998, KBSR has been dedicated to preventing birth defects through ensuring timely, complete, and accurate birth defects surveillance; referring children with birth defects to appropriate services; providing data for research studies after appropriate review; and implementing birth defects prevention programs in Kentucky. KBSR collects data on Kentucky residents from birth to age five who are diagnosed with a structural birth defect or disabling condition as specified in Kentucky Administrative Regulations (see 902 KAR 19:010 in Appendix 2), using data from vital records, acute care and birthing hospitals, laboratories, other state agency surveillance systems, and genetics clinics.

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 activities:

### Surveillance

- Create and maintain a registry of birth defects in Kentucky;
- Analyze the patterns of birth defects in Kentucky;
- Monitor data for changes in rates through time and geography;
- Evaluate timeliness and quality of data on birth defects ; and
- Compile and disseminate surveillance data.

### Research

- Facilitate research studies to help identify causes of birth defects; and
- Respond to requests for aggregate data after approval from an Institutional Review Board and a signed data sharing agreement.

### Prevention

- Support the education of the general public and health professionals about the causes, surveillance, impact, and prevention of birth defects; and
- Collaborate with partner organizations at the national, state, and local level to assist with their prevention efforts.

### Services

- Refer identified children and their families to appropriate services; and
- Evaluate the referral program.

## Methodology and Data

### Introduction:

KBSR collects information on children from birth to age five who are diagnosed with structural birth defects and disabling conditions as defined in 902 KAR 19:010. These anomalies are defined by specific International Classification of Disease (ICD) diagnosis codes. A list of these ICD-9 codes and conditions is provided in Appendix 2. However, in this report, the term “birth defects” is defined as all conditions represented by ICD-9 codes ranging from 740.0 to 759.9. As mentioned in a previous section, this group of conditions includes some that have few clinical consequences as well as some that are transient, meaning that they will naturally resolve without medical intervention. Therefore, due to differences in definitions, the counts and rates of all birth defects used in this report may not be comparable to information distributed by other state or national entities.

KBSR is a passive surveillance system; data is reported to KBSR from several sources, but KBSR staff members do not attempt to actively identify cases from other sources. Currently, KBSR receives data from nine sources: Kentucky Hospital Association (KHA), live birth certificate, death certificate, stillbirth certificate, Critical Congenital Heart Defect (CCHD) surveillance, Neonatal Abstinence Syndrome (NAS) surveillance, University of Kentucky (UK) genetics clinics, University of Louisville (U of L) genetics clinics, and genetics laboratories.

### Reporting:

Reporting is required from acute care hospitals, birthing centers, and medical laboratories licensed in Kentucky. KBSR contracts with KHA to report all hospital discharge data on children with conditions monitored by KBSR. This data is provided in an electronic format on a quarterly basis and includes medical information as well as identifiers used for linkage with vital records and other sources. Additionally, KBSR receives data from two medical laboratories on a quarterly basis. This data contains specific laboratory results for genetic testing along with identifiers for linkage.

Hospital outpatient reporting is voluntary. Currently, the UK hospital and the U of L hospital report on cases seen at pediatric genetics or developmental clinics. Both facilities have onsite as well as outpatient clinics and report on all patients. Demographic information, identifiers, and medical diagnoses are provided quarterly in an electronic format.

The Office of Vital Statistics (OVS) provides data on children with birth defects indicated on the Certificate of Live Birth, Still Birth, or Death. Additionally, KBSR has a Memorandum of Understanding (MOU) with OVS so that KBSR cases can be linked with their relevant vital records, regardless of the data source for the birth defect. This linkage is a crucial source of information about demographics and risk factors during pregnancy, and it also allows for tracking survival and outcomes among children with birth defects. This data is received twice monthly and is uploaded electronically into the KBSR database.

Additionally, KBSR receives data from two DPH programs. The first is the Critical Congenital Heart Defects (CCHD) Registry. As of January 1, 2014, CCHD screening became part of the mandatory statewide newborn screening. KBSR receives the results of pulse oximetry screening and (if performed) the echocardiogram. This data is automatically uploaded into KBSR twice a month, reducing the time to case ascertainment.

KBSR also receives data from the Neonatal Abstinence Syndrome (NAS) Surveillance System, which monitors Kentucky residents with symptoms of NAS. This surveillance system was implemented in July 2014. KBSR currently receives this data electronically twice a month, and data are automatically uploaded into the KBSR database. Prenatal exposure to some substances, including alcohol, is linked to specific congenital anomalies. However, there is limited knowledge about the effects of other substance exposures during pregnancy, and the linkage between the NAS Surveillance System and KBSR will facilitate further research on the topic. Please see the Further Directions section (page 48) for more information about NAS and KBSR.

**Confidentiality:**

KBSR operates under strict guidelines of confidentiality per KRS 211.670. All identifying information is strictly safeguarded and is protected by state law from unauthorized release. All staff members of KBSR are employees of the Cabinet for Health and Family Services and have signed the cabinet’s confidentiality policy.

KBSR data is also protected under Kentucky’s Personal Information Security and Breach Investigation Procedures and Practices Act (KRS 61.931-.934), which secures and protects any personal information including names, biometric markers, account numbers, Social Security Numbers, state-issued identification numbers, federally-issued identification numbers, and individually identifiable information as defined by the Health Insurance Portability and Accountability Act (HIPAA).

KBSR staff take extreme caution in protecting the confidentiality of all data received. Specific actions include: 1.) ensuring that electronic reporting to KBSR is sent securely through the cabinet’s secure interface, 2.) the web-based surveillance system is housed on the cabinet’s secure servers, and 3.) hard copy reports that include protected health information (PHI) are locked in a filing cabinet.

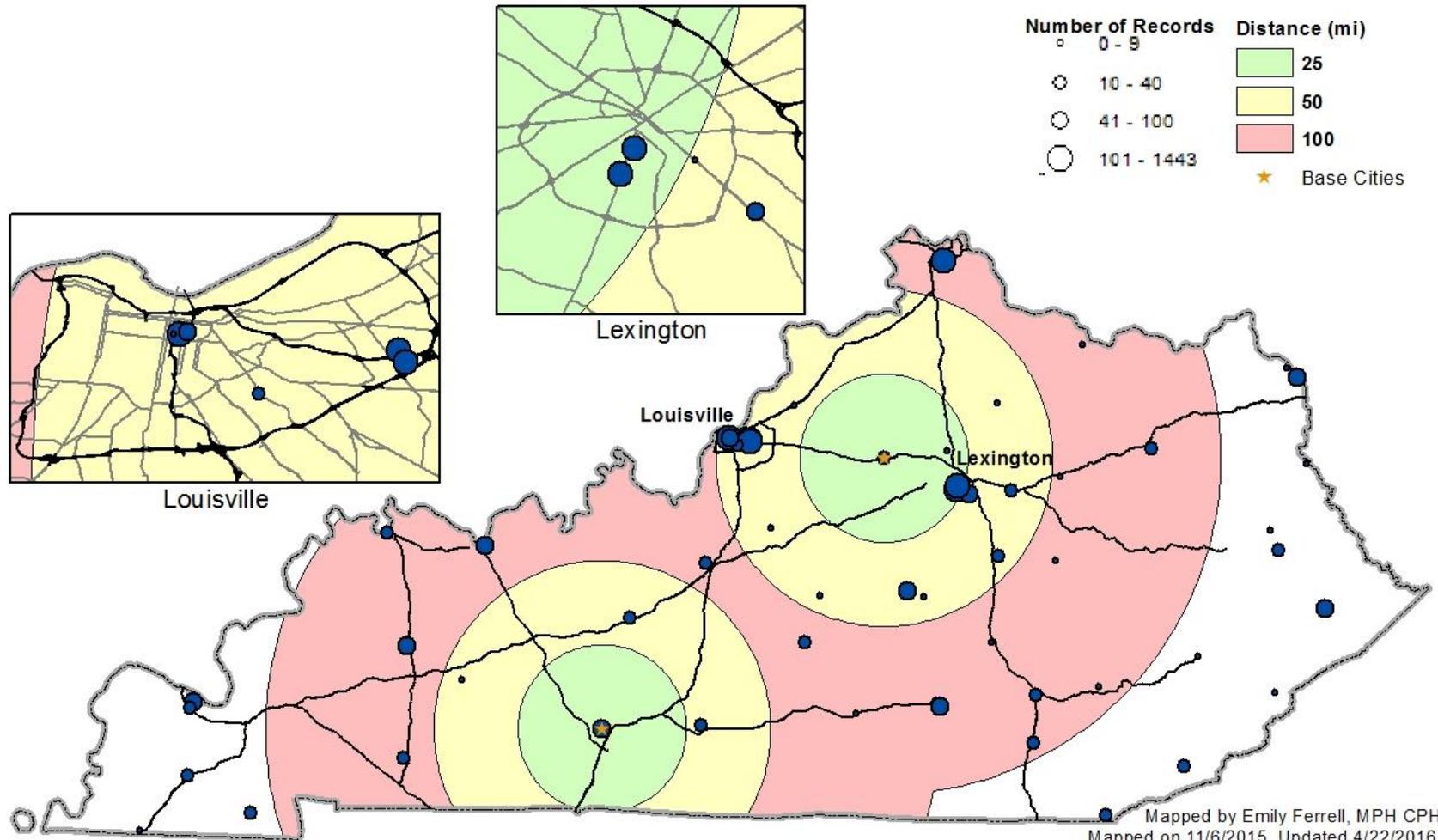
**Abstraction:**

To confirm hospital discharge and Vital Statistics records gathered by passive surveillance, abstractors conduct a broad review of medical records to verify the birth defect or diagnosis and to ensure that all birth defects are ascertained. Abstraction of 44 conditions (Appendix 3) from hospitalizations occurring in 2011-2014 was completed in Spring-Fall 2015. Approximately 3,620 cases were reviewed with information from 4,749 charts at 56 facilities in Kentucky (more than one chart could be reviewed per case). A map indicating the number of charts reviewed per facility and the facility’s distance from KBSR staff base locations is located on the next page. This map will be used to plan the logistics of chart abstraction to maximize efficiency and reduce resource expenditures. In August 2015, KBSR staff held an internal meeting to update the list of conditions abstracted to better align with the reportable conditions included on the National Birth Defects Prevention Network (NBDPN) Annual Report. Starting in Spring 2016, KBSR will abstract 50 conditions on a quarterly basis, including 45 of the 50 conditions reported to NBDPN.

# KBSR Chart Abstraction by Hospital

Spring-Fall 2015

Number of Records Included = 4,749



Mapped by Emily Ferrell, MPH CPH  
Mapped on 11/6/2015. Updated 4/22/2016.

Data from KBSR program and Commonwealth Office of Technology Division of Geographic Information

**Database:**

Prior to 2015, KBSR data was housed in a Microsoft Access database, which allowed data from each source to be stored in its own table and linked together using a common unique Case ID. In October 2015, KBSR’s web-based surveillance system went into live production. The web-based system allows KBSR to link data from multiple data sources to provide a comprehensive view of each case with a separate web page for information from each data source. The system facilitates the processes of chart abstraction by allowing selected users to edit diagnosis and tracking information. The web-based system enhances the current population-based birth defects surveillance system with features that include accessibility in the field, a complex linkage algorithm, and the capability for system generated and ad hoc reports.

**Analytic Methods:**

Within this report, results are presented for selected defects among live births and stillbirths from 2005-2014, regardless of whether the defect was isolated or syndromic. A case is a child, infant, or fetus with the specified birth defect (defined by ICD-9/ICD-10 codes).

Rate calculations are the estimated prevalence of conditions per 10,000 live births. Birth prevalence is calculated as follows:

$$\frac{\text{Number of cases x 10,000}}{\text{Total number of live births}}$$

Although this report 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.

**Data Suppression and Stability:**

In all analyses of KBSR data, rates are suppressed if a cell count is <5 to ensure the protection of sensitive data in small populations and prevent 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 suppressed. In addition, rates that are based on cells with counts ranging from 5-19 are noted as being unstable. The numbers are very small, represent rare conditions, and should be interpreted with caution.

When rates are graphed over time, 3-year rolling averages are used. This method increases the case counts per data point, which can improve the stability of the estimates. The result is that the rates are somewhat smoothed and less likely to be skewed by outliers. An example calculation for the years 2005-2007 is below:

$$\frac{(\text{Number of cases in 2005} + \text{Number of cases in 2006} + \text{Number of cases in 2007}) \times 10,000}{(\text{Total number of live births in 2005} + \text{Total number of live births in 2006} + \text{Total number of live births in 2007})}$$

**Limitations:**

One of the limitations of KBSR is that it is a passive surveillance system, which can affect the timeliness of data reporting. With some data reported on a quarterly basis, the lag time can be significant (up to 6 months after the diagnosis was made). However, as reporting in KBSR continues through age 5 the effect of this limitation is lessened as all diagnoses should be reported to KBSR within that timeframe.

Another limitation of KBSR is the lack of information on pregnancy outcomes other than live births and stillbirths, which are included in the database if they indicate a birth defect or a high-risk maternal condition. In Kentucky, stillbirth certificates are only issued for fetal deaths of 20 completed weeks gestation or more (calculated from the date the last normal menstrual period began to the date of delivery) or in which the fetus weighs 350 grams or more.<sup>5,6,7</sup> Therefore, elective terminations and fetal demises prior to 20 completed weeks gestation or in which the fetus weighs less than 350 grams are not included in KBSR.

Also, cases that are identified at facilities outside of Kentucky or at outpatient facilities that do not report to KBSR (such as prenatal diagnostic facilities and private physicians' offices) are not available to KBSR. This issue is of particular concern in areas near state borders, particularly when there is a large city with specialized pediatric care in a neighboring state. To address this limitation, KBSR plans to work with large hospitals in neighboring states to implement data sharing agreements.

As with most birth defects registries, one concern is that the data are collected from medical records and are therefore subject to differences in clinical practice. The process of chart abstraction helps to minimize the inconsistencies, but abstraction only occurs for certain conditions and does not guarantee complete uniformity in diagnostic coding.

A final limitation of the data in this report is that over the course of 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 ICD codes used for specific diagnoses (such as gastroschisis and omphalocele), the type of coding used (some sources began reporting ICD-10 codes), 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; however, using a 3-year rolling average for rates over time (as previously explained in the Data Suppression and Stability section on page 10) makes the rates more comparable.

## Pregnancy in Kentucky

When examining the occurrence of birth defects, it is important to consider the contextual factors of the pregnancy, including maternal demographics and health concerns. Specific birth defects occur more often in some racial or ethnic groups.<sup>8</sup> Some health behaviors (such as smoking, drinking, and drug use) and health conditions (such as diabetes and hypertension) are associated with an increased risk of specific birth defects.<sup>9</sup> Identifying these at-risk groups could allow for the development of targeted prevention materials and interventions with the goal of reducing birth defects. The factors presented in this section are categorized as demographic (race/ethnicity, maternal age, and maternal education), health-related (diabetes, hypertension, smoking, body mass index, and prenatal care), or gestational age. Each of the birth defects highlighted in this report was analyzed against these factors; results are not presented unless a trend was noteworthy.

### Demographic Factors

Table 1: Demographic Characteristics of Mothers Having a Child With a Birth Defect and All Mothers, Kentucky Residents, 2005-2014

		KBSR 48,862	Kentucky 568,248
Race/Ethnicity	Non-Hispanic White	72.6%	83.2%
	Non-Hispanic Black	15.8%	8.8%
	Hispanic	5.9%	4.8%
	Other/Unknown	5.9%	3.2%
Maternal Age	<20	11.3%	11.4%
	20-24	28.9%	29.3%
	25-29	28.0%	28.9%
	30-34	19.3%	19.7%
	35-39	8.5%	8.0%
	40+	2.1%	1.9%
	Unknown	1.9%	0.8%
Education	<High School	19.4%	18.3%
	High School	27.2%	27.6%
	>High School	50.1%	52.8%
	Unknown	3.2%	1.3%

2005-2014 live births. Kentucky residents only.

Data from Kentucky Birth Surveillance Registry and Kentucky Live Birth Certificates.

2012-2014 data are preliminary and numbers may change.

KBSR births include all cases diagnosed with an ICD-9 code between 740.0-759.9.

N (KBSR)=48,862. N (Kentucky)=568,248.

**Race and Ethnicity:** Every year, over 55,000 Kentuckians are born to a diversifying population. According to the combined data from 2005-2014, Kentucky's population of birthing mothers is predominately Non-Hispanic White (83.2%). However, about 9% of this population is Non-Hispanic Black, about 5% is Hispanic, and about 2% is another or unknown race. Among mothers who have a child with a birth defect, minority populations make up a larger percent of the population. The increase in Non-Hispanic Black mothers can be partially attributed to the higher prevalence of preterm birth in that population. Atrial septal defect (ICD-9 745.5, also known as patent foramen ovale) occurs in much higher rates among preterm infants, although many of the cases are transient.<sup>10</sup> On average, more than 1,100 cases of atrial septal defects are reported to KBSR every year, and Non-Hispanic Blacks have 1.85 times higher risk of having the condition than Non-Hispanic Whites. Therefore, the higher percent of Non-Hispanic Blacks among mothers of children with a birth defect may be partly due to a transient condition associated with preterm birth.

**Maternal Age:** There are certain birth defects associated with both teenage pregnancies (for example, gastroschisis) and advanced maternal age (such as Down Syndrome), which makes maternal age an important variable. In Kentucky, the majority of births (approximately 60%) are to women in their 20s. Another 20% of births are to women in their early 30s. Approximately 10% of births are to women of advanced maternal age, defined as 35 years or older. An additional 10% of all pregnancies are among teenagers. Nationally, and in Kentucky, the rate of teenage pregnancies is steadily decreasing (data not shown).<sup>11</sup> The distribution of maternal age at time of birth is very similar for mothers of children with a birth defect, compared to all mothers in Kentucky's birth population. This similarity in the two groups could be due to the low prevalence of birth defects associated with maternal age.

**Education:** One reason that mother's education is considered as a factor in certain birth defects is its correlation with high risk behaviors such as substance abuse and smoking. Also, educational attainment is often used as a proxy measure for income. Women who have higher incomes might be more likely to access prenatal care, have better access to healthy foods and vitamins, or be better able to afford treatments and interventions for a child with a birth defect. Of mothers in Kentucky, nearly 20% do not have a high school degree or equivalent credential; over 25% of mothers have a high school degree or equivalent credential but no additional education; and about half of the birth population has at least some college or additional education. However, in this dataset, the differences in educational attainment among mothers of children with birth defects and all Kentucky mothers are minimal.

## Maternal Health Factors

Table 2: Selected Health Characteristics of Mothers Having a Child With a Birth Defect and All Mothers, Kentucky Residents, 2005-2014

		KBSR 48,862	Kentucky 568,248
Diabetes	Pre-existing	1.8%	0.8%
	Gestational	5.2%	4.6%
	None	90.6%	91.5%
	Unknown	2.3%	3.1%
Hypertension	Pre-existing	1.9%	1.4%
	Gestational	7.0%	5.5%
	None	88.8%	90.0%
	Unknown	2.3%	3.1%
Smoking	Smoker	22.0%	23.3%
	Heavy Smoker	8.0%	6.4%
	Non-Heavy Smoker	14.0%	16.9%
	Non-Smoker	76.0%	76.3%
	Unknown	2.0%	0.5%
BMI	Underweight	4.4%	4.7%
	Normal	38.9%	42.3%
	Overweight	23.3%	23.8%
	Obese	28.5%	26.1%
	Unknown	4.9%	3.1%
Prenatal Care	Inadequate	4.3%	4.6%
	Intermediate	17.7%	20.3%
	Adequate	35.1%	39.4%
	Adequate Plus	35.2%	31.0%
	Unknown	7.7%	4.8%

2005-2014 live births. Kentucky residents only.

Data from Kentucky Birth Surveillance Registry and Kentucky Live Birth Certificates.

2012-2014 data are preliminary.

KBSR births include all cases diagnosed with an ICD-9 code between 740.0-759.9.

N (KBSR)=48,862. N (Kentucky)=568,248.

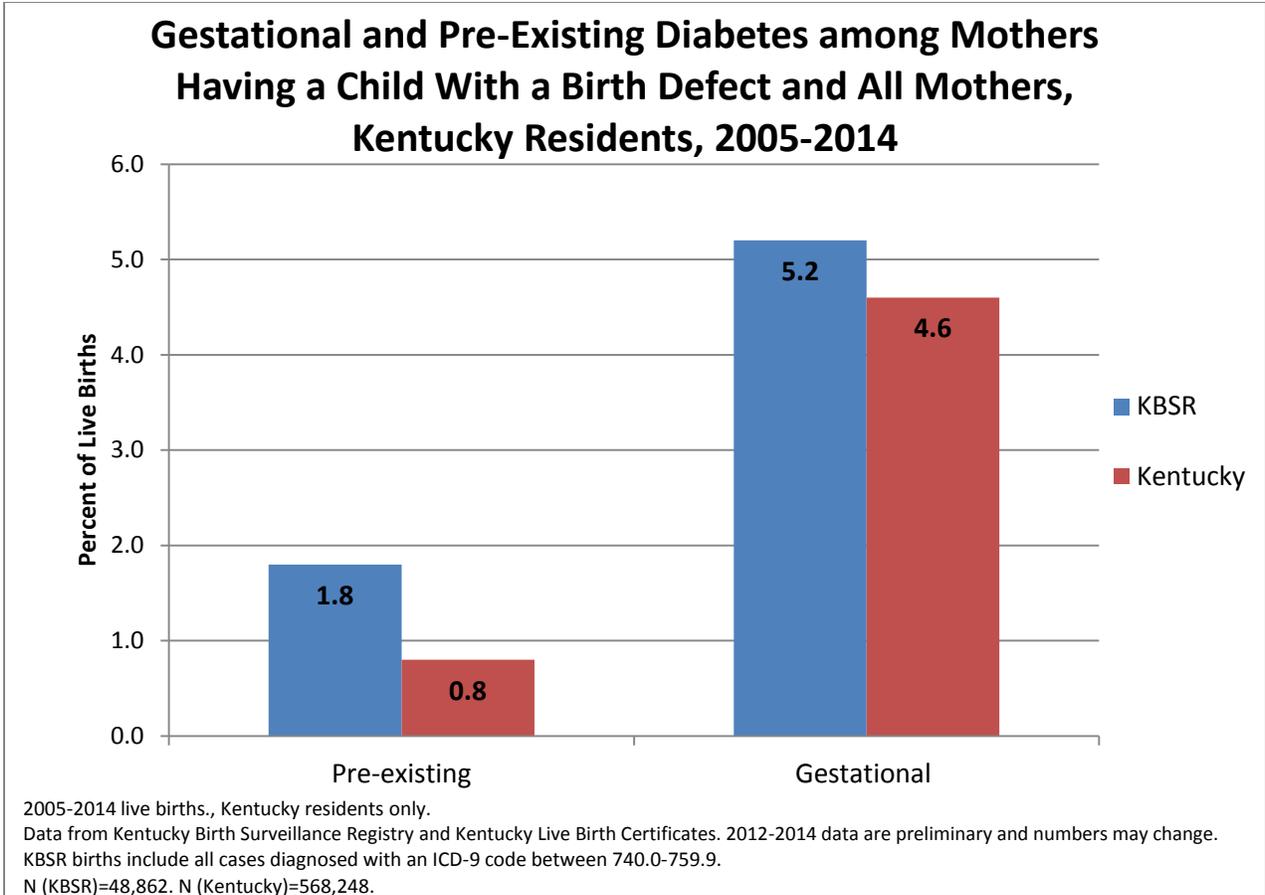
Standard definitions are used for smoking intensity. Heavy smoking is defined as >10 cigarettes per day during the last trimester of pregnancy. Non-heavy smoking is defined as 10 or fewer cigarettes per day during the last trimester of pregnancy.

BMI is based on mother's reported pre-pregnancy weight and height.

Prenatal care categories based on Kotelchuck index, which combines the timing of initiation of prenatal care with the number of prenatal visits (adjusted for gestational age).

**Diabetes:** Uncontrolled diabetes can cause an increased risk of birth defects, particularly congenital heart defects.<sup>12</sup> In Kentucky, 5.4% of all mothers have diabetes when they deliver (4.6% have gestational diabetes and 0.8% have diabetes prior to pregnancy). Among mothers of children with birth defects, 7% have diabetes when they deliver. Although the percent of mothers with gestational diabetes is slightly higher, the main difference is in the percent with diabetes prior to pregnancy (1.8% versus 0.8%).

Graph 1: Gestational and Pre-Existing Diabetes among Mothers Having a Child With a Birth Defect and All Mothers, Kentucky Residents, 2005-2014



**Hypertension:** Like all chronic or pregnancy-related health conditions, hypertension should be managed during pregnancy to promote optimal outcomes for mother and baby. Hypertension during pregnancy is linked to several adverse pregnancy outcomes including premature birth, low birth weight, and stillbirth.<sup>13</sup> Women with hypertension should have a conversation with their healthcare providers about medication use during pregnancy.<sup>4</sup> In Kentucky, 6.9% of all mothers have hypertension when they deliver (5.5% have gestational hypertension and 1.4% have hypertension prior to pregnancy). Among mothers of children with birth defects, 8.9% have hypertension at delivery (7.0% have gestational hypertension and 1.9% have hypertension prior to pregnancy).

**Smoking:** Smoking during pregnancy increases the risk of having a baby with certain birth defects, including heart defects and orofacial clefts.<sup>14</sup> It also may cause low birth weight or prematurity, placenta previa (when the placenta covers the cervix, causing severe bleeding before or during delivery), or placental abruptions.<sup>15</sup> Smoking harms the health of the baby and mother by reducing the amount of oxygen they breathe and exposing them to harmful chemicals. As more than 1 in 5 women in Kentucky smoke during pregnancy, smoking is one of the most common and significant risk factors for adverse pregnancy outcomes in Kentucky. There is a dose-response relationship between exposure to cigarette smoke and adverse pregnancy outcomes. While it is ideal for a woman to quit smoking during pregnancy, there could also be some benefit to reducing the number of cigarettes smoked. Smoking intensity was categorized according to standard practice as either heavy (mother smoked >10 cigarettes per day during the last trimester of pregnancy) or non-heavy (10 or fewer cigarettes per day during the last trimester of pregnancy). There was little difference in rates of smoking during pregnancy among mothers of children with birth defects versus Kentucky mothers as a whole; in both groups, there were more light smokers than heavy smokers. However, mothers of children with birth defects were more likely to be heavy smokers (8.0% and 6.4%, respectively). A section of this report (Fetuses Affected by Substance Exposure, page 38) gives further information about smoking and other substance exposures during pregnancy.

**Pre-Pregnancy Body Mass Index (BMI):** Overweight and obesity during pregnancy are associated with some birth defects, such as neural tube defects, and can contribute to complications of pregnancy and delivery.<sup>16</sup> Maternal BMI has caused national concern and is a particular issue in Kentucky. About one-half of mothers in Kentucky are overweight or obese prior to conception; approximately 10% of mothers are underweight; and about 40% of mothers fall into the normal or healthy BMI range. There are no significant differences in BMI between mothers of children with birth defects and all mothers in Kentucky

**Prenatal Care:** Access to early and adequate prenatal care is an important aspect of birth defects prevention. Prenatal care provides not only medical interventions and diagnostics, but also serves to educate expectant mothers on lifestyle factors and risk reduction. In this report, prenatal care is categorized as follows: inadequate, intermediate, adequate, adequate plus, or missing (incomplete on birth certificate). These categories were determined using the Kotelchuck index for adequacy of prenatal care, which combines the timing of initiation of prenatal care with the number of prenatal visits (adjusted for gestational age).<sup>17</sup> Among mothers in Kentucky and mothers of children with birth defects, less than 5% had inadequate prenatal care and about 20% had intermediate prenatal care. For all mothers in Kentucky and for mothers of children with birth defects, approximately 70% had adequate or adequate plus prenatal care. The main difference between the two is in the distributions: 35.1% of mothers of children with birth defects had adequate care and 35.2% had adequate plus care, while among Kentucky mothers the respective percentages were 39.4% and 31.0%. This difference could indicate that birth defects were diagnosed prenatally, and these women had more prenatal visits than they would have had otherwise. Another possible factor is that women who have a comorbid condition, such as pre-existing diabetes or hypertension, receive more prenatal care visits and are also at higher risk of having a child with a birth defect.

## Gestational Age

Gestational age can be categorized as early preterm (less than 32 weeks), preterm (32-33 weeks), late preterm (34-36 weeks), early term (37-38 weeks), and term (greater than or equal to 39 weeks). There is a relationship between gestational age and birth defects. Birth defects that are incompatible with life often result in preterm deliveries. Premature (less than term) infants are more likely to have certain birth defects, such as transient heart defects that resolve with time.<sup>10</sup> Premature infants are also more likely to die from a birth defect, as opposed to term infants. Prenatal diagnosis with certain birth defects could result in a medically-indicated early delivery. The percent of children with birth defects that were born at less than 37 weeks is nearly twice that of the birth population (21.8% versus 11.7%). The main difference is in births that occurred at less than 32 weeks gestational age (early preterm deliveries). Approximately 7% of children with birth defects fall into this category, while only 1.7% of the Kentucky population does. While the percentages of early term births are comparable in both populations, a smaller percent of children with birth defects were born at full term (46.2% versus 55.9%).

Table 3: Gestational Age Distribution among Births in KBSR and All Live Births, Kentucky Residents, 2005-2014

	KBSR 48,862	Kentucky 568,248
Early Preterm (<32 wks)	7.0%	1.7%
Preterm (32-33 wks)	3.0%	1.3%
Late Preterm (34-36 wks)	11.8%	8.7%
Early Term (37-38 wks)	30.2%	32.3%
Term (39+ wks)	46.2%	55.9%
Unknown	1.9%	0.2%

2005-2014 live births. Kentucky residents only.

Data from Kentucky Birth Surveillance Registry and Kentucky Live Birth Certificates.

2012-2014 data are preliminary and numbers may change.

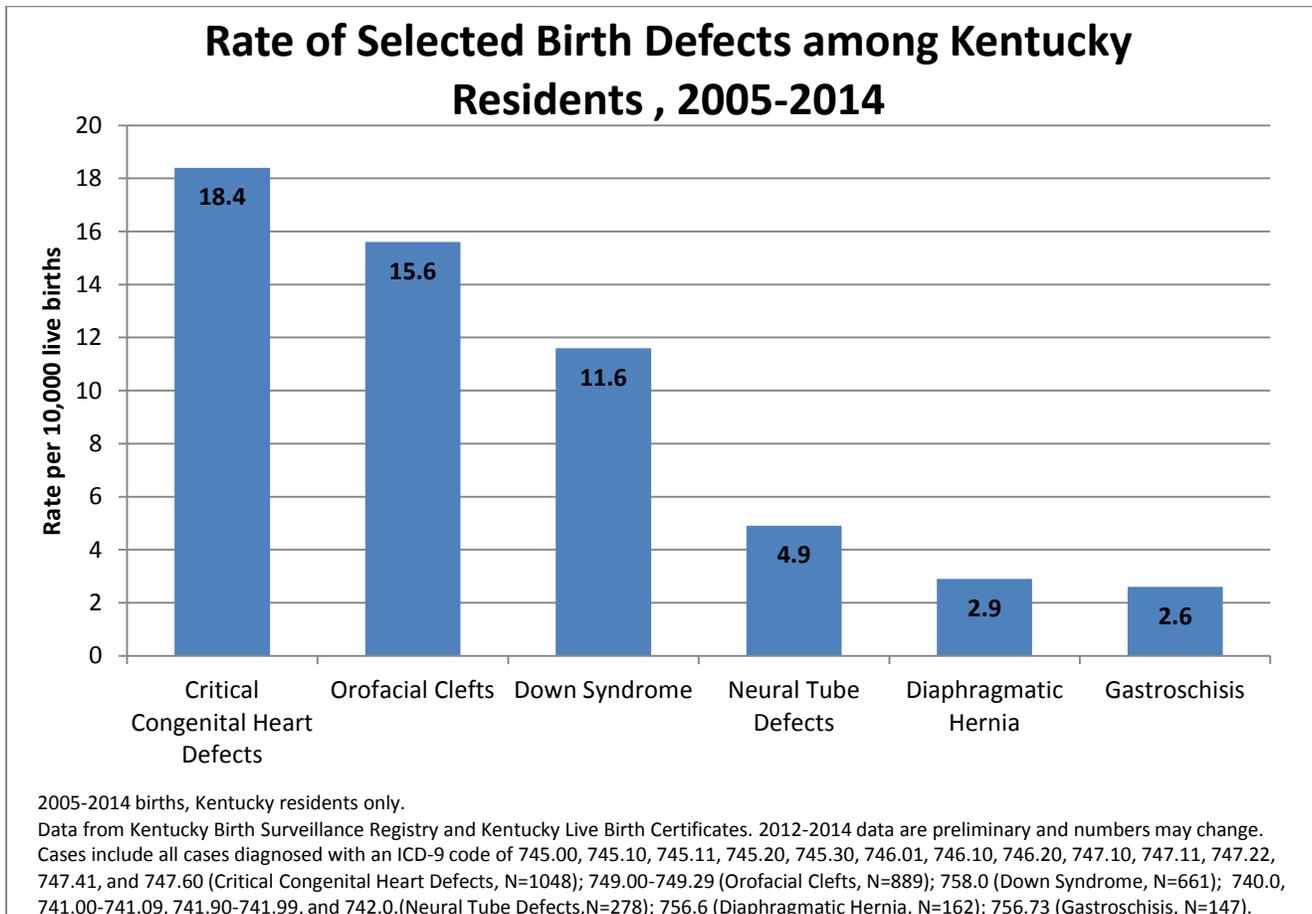
KBSR births include all cases diagnosed with an ICD-9 code between 740.0-759.9.

N (KBSR)=48,862. N (Kentucky)=568,248.

## Selected Birth Defects

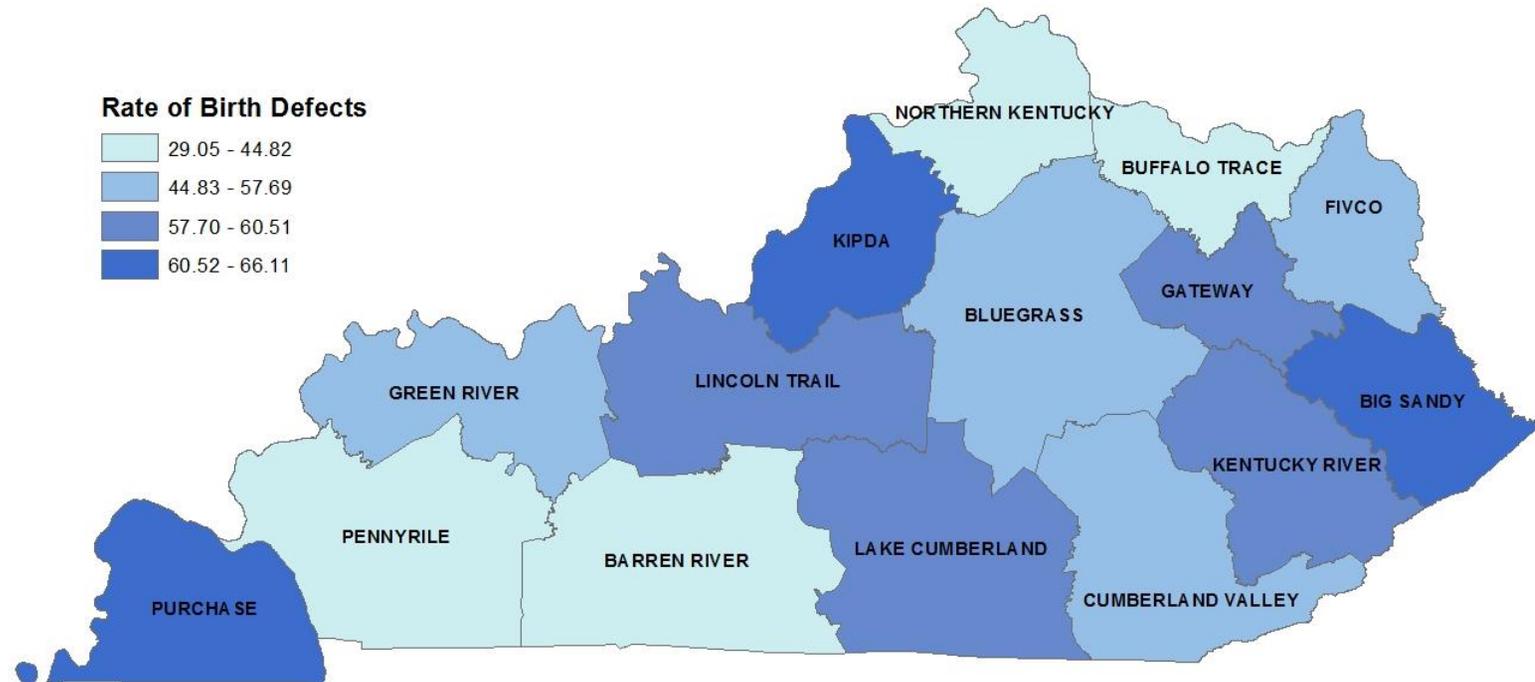
The table below presents some common major birth defects in Kentucky, which will be highlighted in this report. Some are as common as 18.4 cases per 10,000 live births, which is the equivalent of nearly 2% of births. These conditions may require surgery or other treatments and can seriously affect the growth, development, and survival of a child. For more information on the birth defects in the table below, please see the corresponding section of the report.

Graph 2: Rate of Selected Birth Defects among Kentucky Residents, 2005-2014



The map on the following page reveals the rate of these birth defects in each of the Area Development Districts (ADDs) of Kentucky based on child’s county of residence. The rates range from 29 to 66 cases per 10,000 live births. Purchase, Big Sandy, and KIPDA ADDs had the highest rates and were more than twice as high as the rate in the lowest district (Northern Kentucky). Knowledge of geographic patterns of birth defects prevalence can be used to plan prevention activities and services to maximize their effectiveness and reach at-risk populations. The areas of Kentucky with health disparities are the Appalachian region in the east and the Mississippi Delta region in the far west. One possible explanation is that Kentucky children living near Cincinnati or Nashville, might seek specialized pediatric care out-of-state and would be under-represented in KBSR data.

# Rate of Selected Major Birth Defects by ADD, Kentucky, 2005-2014



February 25, 2016

Data Source: Kentucky Birth Surveillance Registry, Kentucky Live Birth Certificates, Kentucky Stillbirth Certificates

Cases definition: ICD-9 codes 745.00, 745.10, 745.11, 745.20, 745.30, 746.01, 746.10, 746.20, 747.10, 747.11, 747.22, 747.41, and 747.60 (Critical Congenital Heart Defects); 749.00-749.29 (Orofacial Clefts); 758.0 (Down Syndrome); 740.0, 741.00-741.09, 741.90-741.99, and 742.0 (Neural Tube Defects); 756.6 (Diaphragmatic Hernia); 756.73 (Gastroschisis)

Shapefiles from Kentucky Geography Network

Prepared by Emily Ferrell, MPH CPH

Notes: Kentucky residents only. Rates are per 10,000 live births. 2012-2014 data are preliminary and numbers may change.

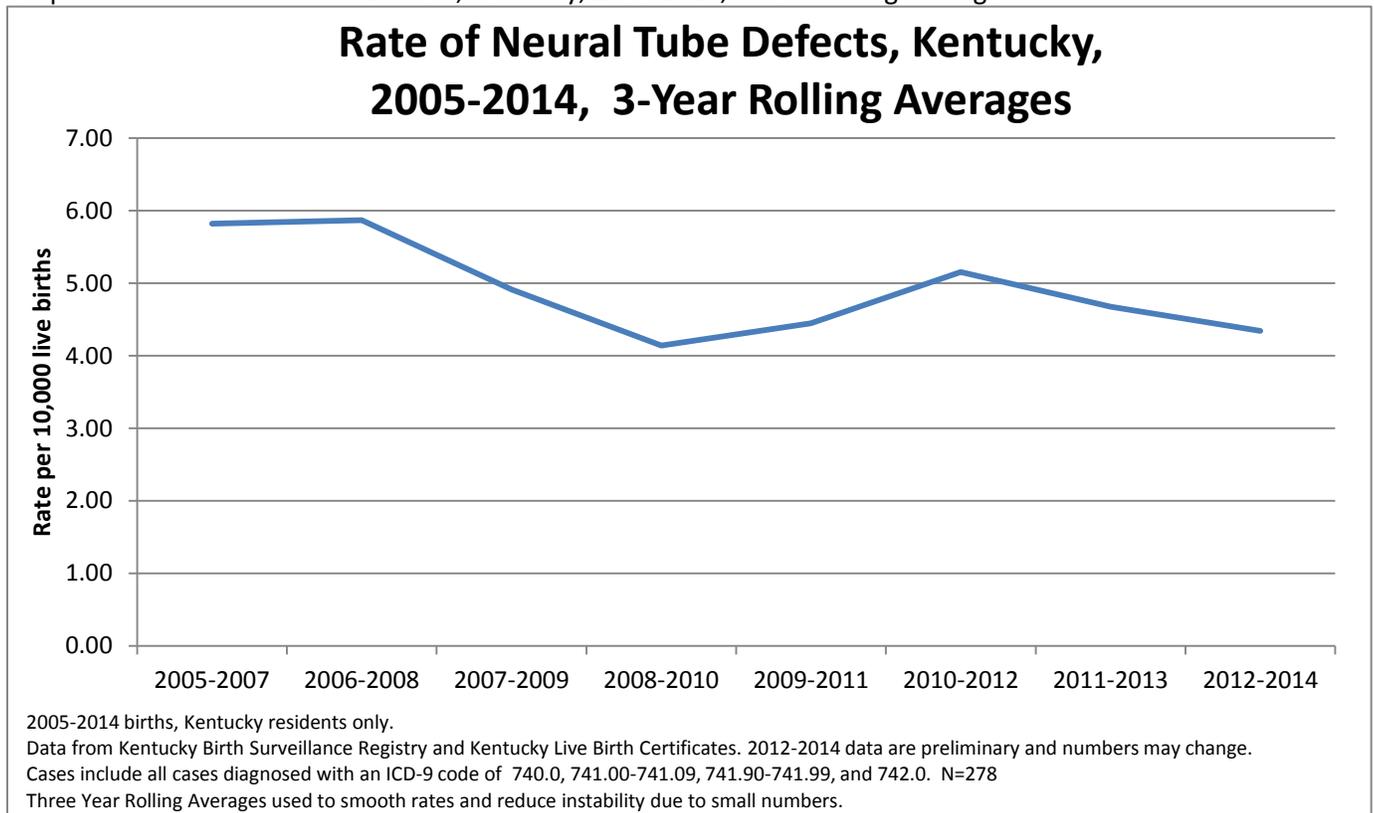
## Neural Tube Defects

Neural tube defects (NTDs) are a group of birth defects that affect the central nervous system. The neural tube usually closes around the third or fourth week of pregnancy.<sup>18</sup> For babies with NTDs, the tube has not closed properly.<sup>19</sup>

According to 2005-2014 KBSR data, about 28 children are born with an NTD every year. This corresponds to a rate of 4.9 cases per 10,000 live births. Using data from 2009-2011, estimates for NTD (anencephaly and spina bifida only) prevalence in the U.S. vary from 5.5 to 6.5 cases per 10,000 live births, depending on surveillance methodology.<sup>20</sup> This corresponds to anywhere from 2,203-2,604 new cases annually nationwide.<sup>20</sup>

The rate of NTDs is declining in Kentucky (see Graph 3) as well as in the U.S. (not shown). Much of this decline can be attributed to the fortification of grains and grain products (including wheat flour) with folic acid<sup>19</sup>. In Kentucky, the rate has decreased from 5.8 cases per 10,000 live births in 2005-2007 to 4.4 cases per 10,000 live births in 2012-2014.

Graph 3: Rate of Neural Tube Defects, Kentucky, 2005-2014, 3-Year Rolling Averages



It is believed that most NTDs occur due to a combination of genetic factors and environmental factors (such as not having enough folic acid or vitamins in the diet).<sup>19</sup> Women with certain health problems like diabetes, seizure conditions, or obesity may be at a higher risk of having a child with an NTD.<sup>19</sup> According to KBSR data from 2005-2014, obese women had a 36% higher risk of delivering a baby with an NTD compared to women with a normal pre-pregnancy BMI. Women who have had a previous pregnancy affected by an NTD have a higher risk of having another child with an NTD.<sup>19</sup> Hispanics are at an increased risk for NTDs compared to Non-Hispanic Whites.<sup>8</sup>

Research has shown that intake of folic acid reduces the risk of NTDs.<sup>18</sup> However, people who take folic acid may still have babies with NTDs, and women who do not take folic acid can still have healthy babies.<sup>19</sup>

### ***Public Health Feature: Folic Acid Supplementation***

The Centers for Disease Control estimates that most neural tube defects can be prevented if a woman takes enough folic acid during the first four weeks after conception.<sup>21</sup> As many pregnancies are undetected during those first four weeks, it is important for **all** women of childbearing age to consume folic acid. The current recommendation from the U.S. Preventative Services Task force is 400 micrograms (mcg) of folic acid daily.<sup>22</sup> The August 1991 U.S. Public Health Service guidelines call for women who have had a previous pregnancy affected by a NTD to consume 4 milligrams (4000 mcg) of folic acid daily beginning one month before they start trying to get pregnant and continuing through the first three months of pregnancy.<sup>23</sup>

For over 15 years, KBSR has worked with partner organizations throughout Kentucky to increase awareness of folic acid's role in birth defects prevention. Due in part to the efforts of many partners to make folic acid more easily available (both through grain fortification and through supplements), the rate of NTDs has been decreasing across Kentucky and the nation.

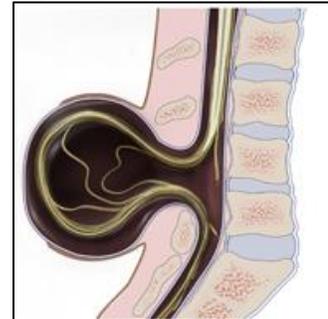
KBSR has worked with the Kentucky Folic Acid and Perinatal Partnership (KFAP) since 1998 to develop, implement, and evaluate folic acid promotion efforts. Recently, KFAP has broadened its focus to include such topics as Folic Acid and Nutrition, Smoking in Pregnancy, and Substance Use in Pregnancy.

The three defects that comprise NTDs are **anencephaly**, **spina bifida**, and **encephalocele**.

**Anencephaly:** Anencephaly occurs when the upper part of the neural tube does not close and the baby's brain and skull fail to form completely.<sup>24</sup> The baby is born without the forebrain (front of the brain) and the cerebrum (area of the brain where thinking and coordination occur). Without a complete brain, a baby's body cannot grow and function.<sup>24</sup> Unfortunately, there is no treatment for children with anencephaly and most affected babies die shortly after birth.<sup>24</sup> Based on KBSR data from 2005-2014, there are about 7 cases of anencephaly annually in Kentucky or about 1.6 cases per 10,000 live births. This number is probably an underestimate as anencephaly is incompatible with life, and cases of stillbirths that do not meet the criteria for a vital record would not be included in KBSR (as previously discussed in the Limitations section on page 11). The estimate for the U.S. is 859 cases annually, which is about 2 cases per 10,000 live births.<sup>25</sup>

**Spina bifida:** Generally, when people speak of spina bifida, they are referring to the most common and also most severe form, called myelomeningocele, although there are several other forms.<sup>19</sup> This condition causes the spinal nerves to bulge through an opening in the back (see Figure 2). The myelomeningocele usually occurs in the lowest part of the spine but can possibly occur at any level.<sup>26</sup> The region of the spine where the myelomeningocele occurs impacts the severity of the symptoms; lesions that are higher along the spine affect more of the nervous system and are more severe.

Figure 2: Myelomeningocele



Spina bifida is the most common permanently disabling birth defect that is compatible with living into adulthood.<sup>26</sup> Complications of spina bifida can include partial or full paralysis of the legs, inability to control bowel or bladder function, obesity, skin breakdown, seizures, eye disorders, and learning disabilities.<sup>26</sup> Based on KBSR data from 2005-2014, there are about 17 cases of spina bifida annually in Kentucky, while there are about 1460 cases in the U.S. annually.<sup>25</sup> In Kentucky, there are about 3 cases per 10,000 live births. In the U.S., there are about 3.5 cases per 10,000 live births.<sup>25</sup>

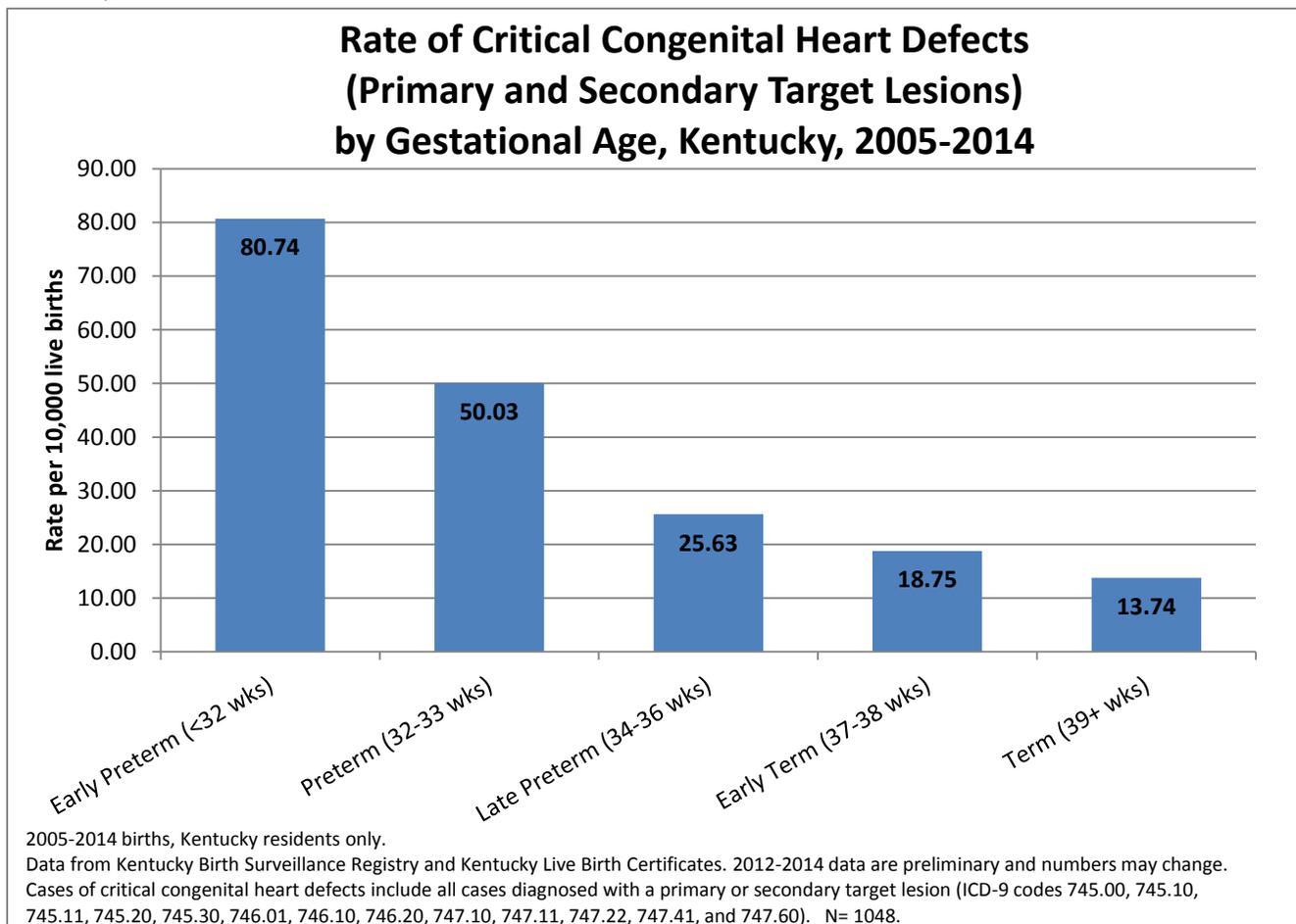
**Encephalocele:** Encephalocele occurs when the neural tube does not close completely, and there is an opening in the midline of the upper part of the skull, the area between the forehead and nose, or the back of the skull.<sup>27</sup> It is described as a sac-like protrusion of the brain and its membranes through an opening in the skull.<sup>27</sup> Sometimes encephalocele is associated with other nervous system problems, motor coordination, developmental or intellectual delays, seizures, and vision problems.<sup>27</sup> Based on KBSR data from 2005-2014, there are about four cases of encephalocele annually in Kentucky. The estimate for the U.S. is 341 cases annually.<sup>25</sup> Both in Kentucky and the U.S., there is less than 1 case per 10,000 live births.<sup>25</sup>

## Congenital Heart Defects:

Congenital heart defects (CHDs) are the most common type of birth defect with about 1 case out of every 100 births.<sup>28</sup> Over 2 million people with CHDs lead active and healthy lives in the U.S. today due to advances in diagnosis and treatment.<sup>28</sup> CHD is a variable category of birth defects when looking at associated outcomes. Some heart defects are transient and might resolve on their own soon after birth. These transient defects are particularly common in premature babies whose hearts did not finish developing prior to delivery.<sup>10</sup> According to KBSR data from 2005-2014 (Graph 4 below), early preterm deliveries have particularly high rates of heart defects, but all categories of preterm births have higher rates when compared to early term and term deliveries.

Some heart defects are so slight that they are not diagnosed until much later in life and do not cause any loss to quality of life. However, other heart defects are much more severe making CHDs the leading cause of birth-defect related deaths.<sup>10</sup> Some defects cause immediate health problems for a baby and require surgery or other interventions within the first year of life.<sup>28</sup> These are known as **critical congenital heart defects (CCHD)**, and they account for 1 in every 4 heart defects. The remainder of this section will be devoted to a discussion of CCHDs.

Graph 4: Rate of Critical Congenital Heart Defects (Primary and Secondary Target Lesions) By Gestational Age, Kentucky, 2005-2014



**Critical Congenital Heart Defects:** Conditions commonly designated as CCHDs 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. These are the heart defects that are most likely to be detected through newborn screening, sometimes called “primary target lesions.”

***Public Health Feature: Pulse Oximetry Screening***

Heart defects can't be detected through a visual examination, so pulse oximetry screening helps health professionals identify heart defects before they cause damage to the baby.<sup>30</sup> Pulse oximetry tells doctors how much oxygen is in a baby's blood. If a baby has a heart defect, the heart might not pump enough oxygenated blood through the baby's body.

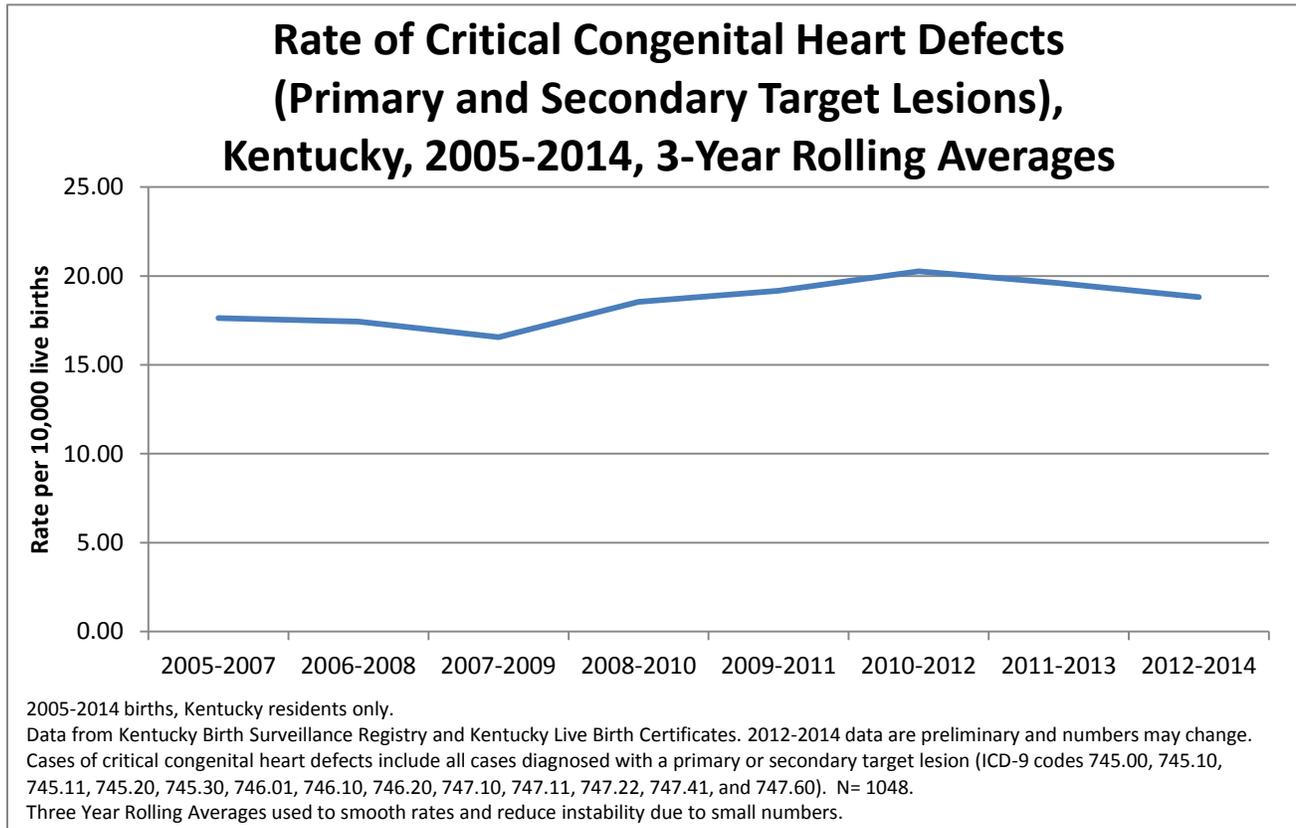
On January 1, 2014, pulse oximetry screening for congenital heart defects became mandatory in Kentucky as part of KRS 214.155.<sup>30</sup> Prior to that time, some hospitals had implemented pulse oximetry screening, but the legislation required that all babies be screened.

Other benefits of pulse oximetry screenings include cost-effectiveness, accuracy, and ease of incorporating into the workflow of the normal newborn nursery.<sup>29</sup> The time required for each screen is about 1 to 5 minutes. A baby with an abnormal pulse oximetry screening result can then be referred for further diagnosis.

In Kentucky and in some other states, Aortic Interruption/Atresia/Hypoplasia, Coarctation/Hypoplasia of Aortic Arch, Double-Outlet Right Ventricle, Ebstein Anomaly, and Single Ventricle are also considered CCHDs due to the severity of these conditions, although they are less likely to be detected by pulse oximetry screening. These are sometimes called “secondary target lesions.”

Rates of CCHDs across geographic areas might not be comparable due to definitions (e.g., inclusion of secondary as well as primary target lesions) as well as differences in screening and reporting methodologies. One study showed that state prevalence estimates of specific CCHDs (2005-2009) were influenced by the practices of newborn screening and birth surveillance registries.<sup>31</sup> Therefore, a U.S. rate is not used as a comparison in this section. Compared to the states in the study, Kentucky's prevalence of specific CCHDs fell within the range of values reported by other state registries. For the overall CCHD rate, Kentucky has about 18.4 cases per 10,000 live births, which is similar to the rate of 17.3 cases per 10,000 live births found in a large Atlanta-based study (both studies included primary and secondary target lesions).<sup>32</sup>

Graph 5: Rate of Critical Congenital Heart Defects (Primary and Secondary Target Lesions), Kentucky, 2005-2014, 3-Year Rolling Averages

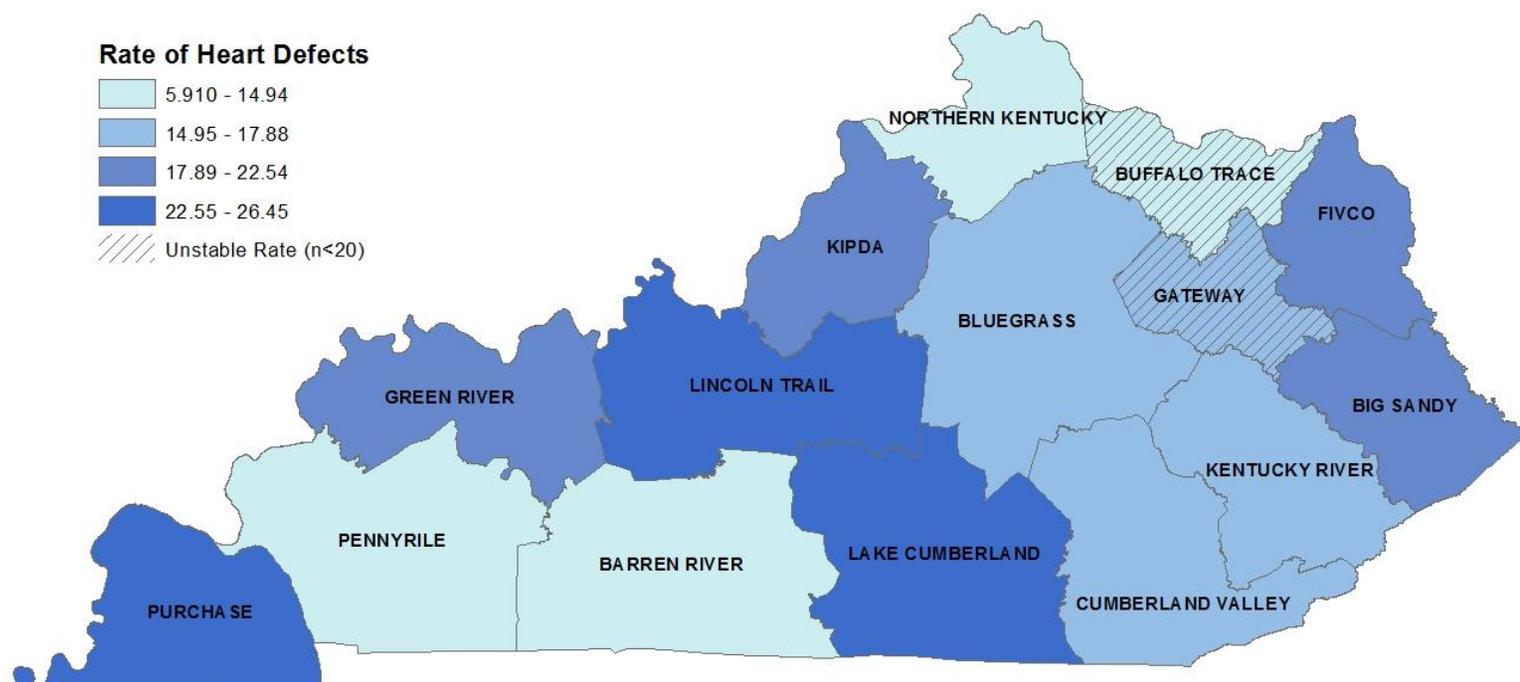


CCHDs (and other CHDs) are thought to be caused by a combination of genes, environmental factors, maternal health conditions, smoking, and maternal medication use during pregnancy.<sup>28</sup> Pre-existing diabetes and obesity have been linked to heart defects in the baby. According to KBSR data from 2005-2014, women with pre-existing diabetes were 4.25 times more likely to have a child with a CCHD than women without diabetes.

The map on the following page displays the rates of CCHDs by Kentucky ADD. The rates vary drastically, from about 6 to 26 cases per 10,000 live births. From this map, it appears that rates are higher in West-Central Kentucky: Green River, Lincoln Trail, KIPDA, and Lake Cumberland ADDs. Rates are also high in the Appalachian ADDs of Big Sandy and FIVCO. The elevated rates in this area may be partially due to increased rates of maternal obesity and smoking (data not shown).

Map 3: Rate of Critical Congenital Heart Defects by Area Development District, Kentucky, 2005-2014

# Rate of Critical Congenital Heart Defects by ADD, Kentucky, 2005-2014



February 25, 2016

Data Source: Kentucky Birth Surveillance Registry, Kentucky Live Birth Certificates, Kentucky Stillbirth Certificates

Cases definition: ICD-9 codes 745.00, 745.10, 745.11, 745.20, 745.30, 746.01, 746.10, 746.20, 747.10, 747.11, 747.22, 747.41, and 747.60.

Shapefiles from Kentucky Geography Network

Prepared by Emily Ferrell, MPH CPH

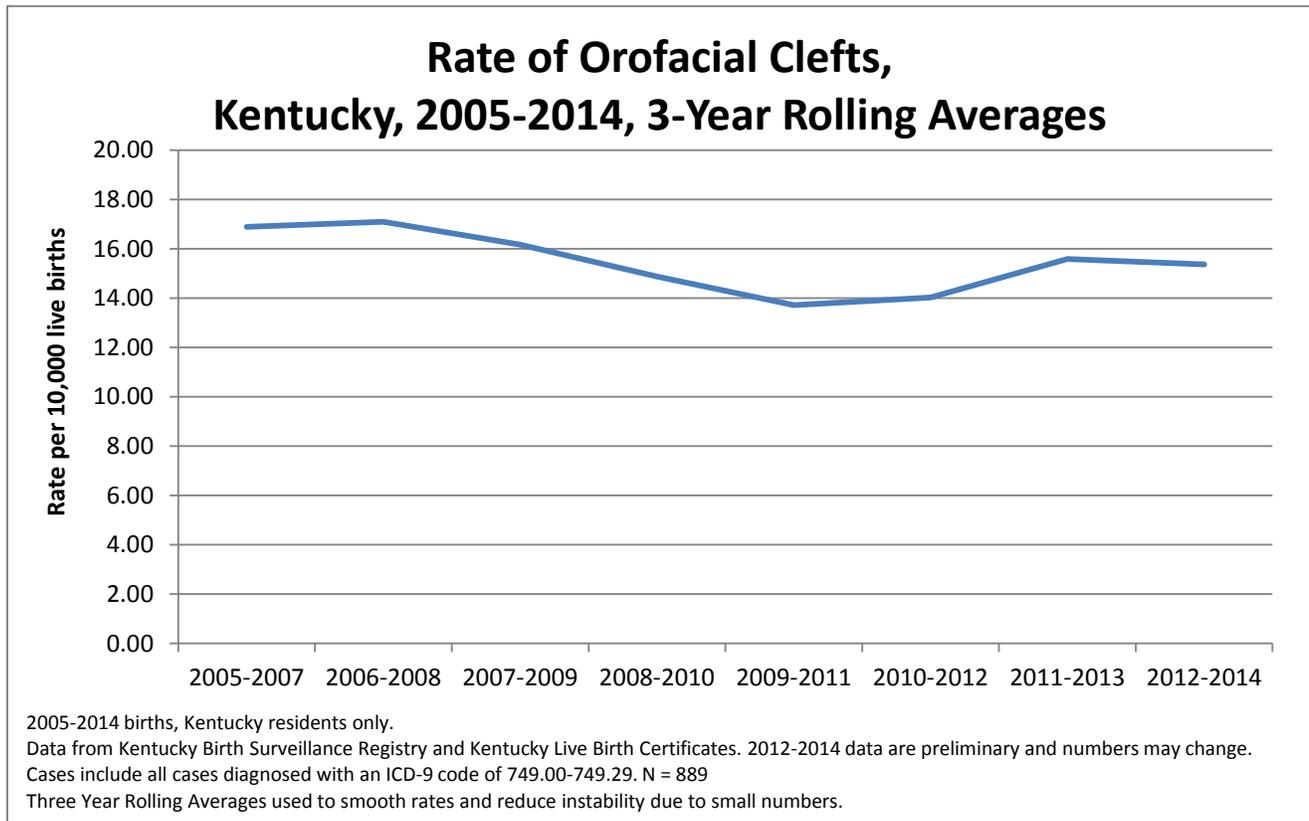
Notes: Kentucky residents only. Rates are per 10,000 live births. 2012-2014 data are preliminary and numbers may change.

## Orofacial Clefts

Cleft lip and cleft palate are birth defects that happen when the tissue that makes up a baby's mouth does not close entirely when it is forming.<sup>33</sup> A baby can have a cleft lip, a cleft palate, or both. All types are known collectively as orofacial clefts, which often occur in the absence of other birth defects (estimated 50-80% of cases).<sup>33</sup> Children with orofacial clefts often have problems with feeding and speech and might also have ear infections, hearing problems, and problems with their teeth.<sup>33</sup>

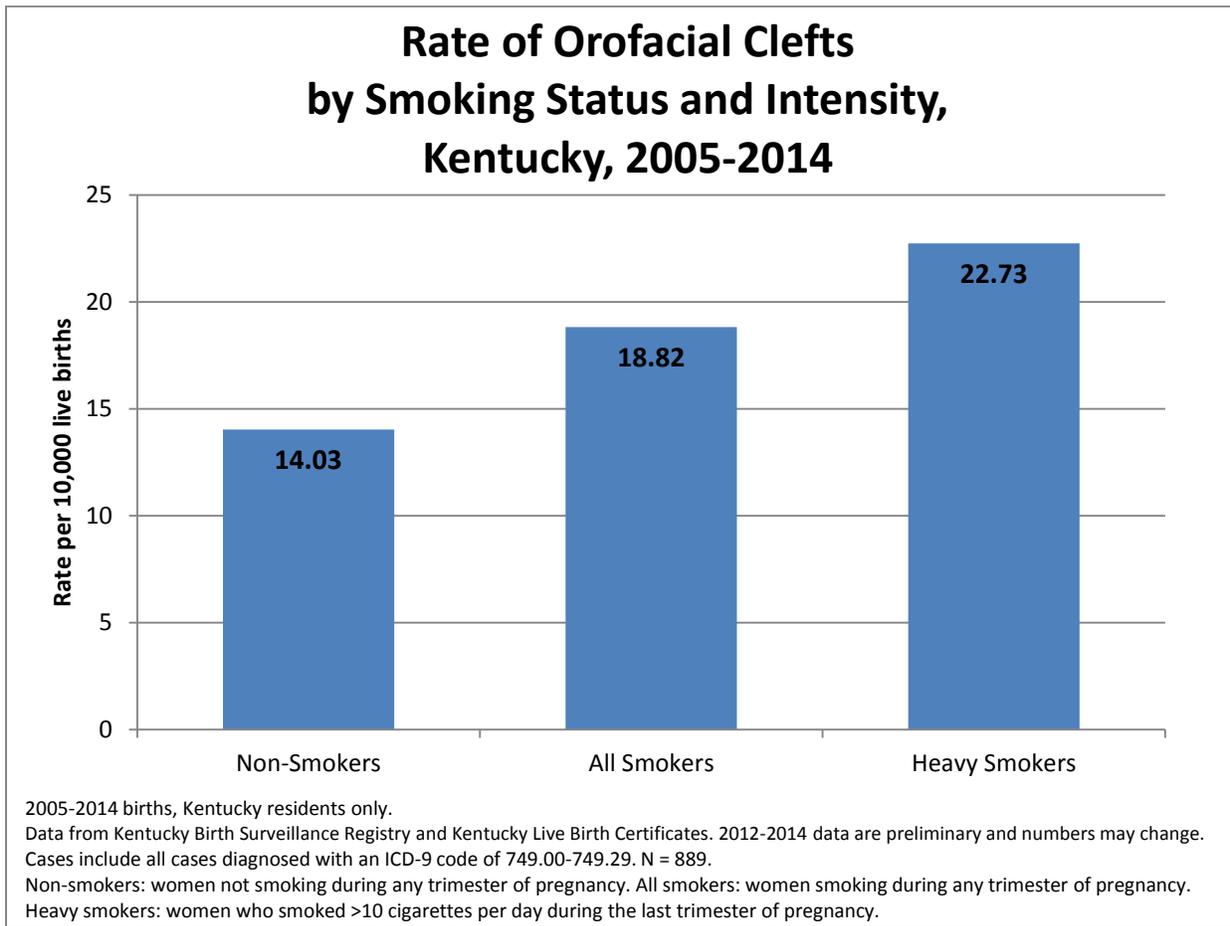
In Kentucky, about 89 children are born every year with an orofacial cleft, which is a rate of 15.6 cases per 10,000 live births. The rate of orofacial clefts has been declining slightly over time (see Graph 6).

Graph 6: Rate of Orofacial Clefts, Kentucky, 2005-2014, 3-Year Rolling Averages



The causes of orofacial clefts are unknown, but several risk factors have been identified, including tobacco use. Studies have shown that women who smoke during pregnancy are more likely to have a child with an orofacial cleft.<sup>33</sup> Compared to non-smokers (women who did not smoke during any trimester of pregnancy), smokers (women who smoked during at least one trimester of pregnancy) were about 35% more likely to have a child with an orofacial cleft according to KBSR data from 2005-2014, regardless of smoking intensity. Heavy smokers (women who smoked >10 cigarettes per day during the last trimester of pregnancy) were over 60% more likely to have a child with a cleft versus non-smokers. This data indicates a trend based on intensity of smoking, with higher risk of orofacial clefts related to greater tobacco exposure.

Graph 7: Rate of Orofacial Clefts by Smoking Status and Intensity, Kentucky, 2005-2014



An additional risk factor for orofacial clefts is maternal diabetes<sup>33</sup>; according to KBSR data from 2005-2014, having pre-existing diabetes doubles the risk of having a child with an orofacial cleft, and gestational diabetes increases the risk by 25%.

Prevalence of orofacial clefts has also been associated with race and ethnicity. According to national studies, orofacial clefts are more common among Non-Hispanic White women compared to Non-Hispanic Black women.<sup>8</sup> KBSR data from 2005-2014 reveal that Non-Hispanic Black women were approximately 35% less likely to have a child with an orofacial cleft, compared to Non-Hispanic White women.

Orofacial Clefts are often categorized as **isolated cleft palate** and **cleft lip with or without cleft palate**. These conditions are described in greater detail below:

**Isolated Cleft Palate:** The roof of the mouth (palate) is formed between the sixth and ninth weeks of pregnancy.<sup>33</sup> A cleft palate happens if the tissue that makes up the roof of the mouth does not join together completely during pregnancy (see Figure 3). For some babies, only part of the palate is open (incomplete cleft palate) while for others, the front and back of the palate is affected (complete cleft palate).<sup>33</sup> Kentucky has about 31 cases of isolated cleft palate per year; in the U.S., there are about 2,651 cases of isolated cleft palate a year.<sup>25</sup> In both Kentucky and the U.S., the rate of isolated cleft palate is about 6.3 cases per 10,000 live births.<sup>25</sup>

Figure 3: Cleft Palate

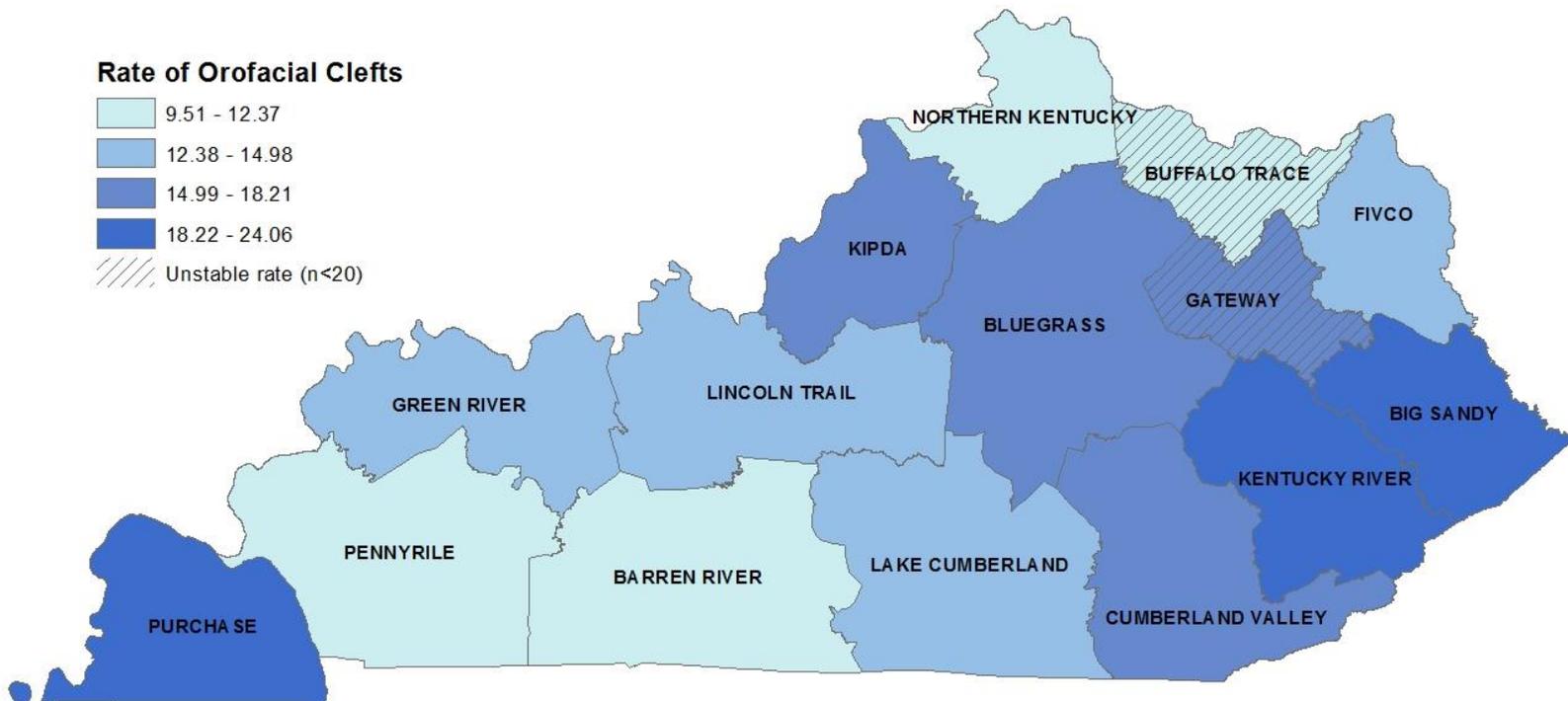


**Cleft Lip with or without Cleft Palate:** The lip forms between the fourth and seventh weeks of pregnancy.<sup>33</sup> A cleft lip happens if the tissue that makes up the lip does not join completely before birth, which results in an opening in the upper lip. The opening in the lip can be a small slit (incomplete cleft lip) or it can be a large opening that goes through the lip into the nose (complete cleft lip). A cleft lip can be on one side of the lip (unilateral cleft lip), both sides of the lip (bilateral cleft lip), or rarely in the middle of the lip. Children with a cleft lip also can have a cleft palate but do not always.<sup>33</sup> Kentucky has about 58 cases of cleft lip (with or without cleft palate) a year; in the U.S., there are about 4,437 cases each year.<sup>25</sup> The Kentucky rate of cleft lip is 10.2 cases per 10,000 live births; in the U.S., the rate is 10.6 cases per 10,000 live births.<sup>25</sup>

The following page contains a map of the rate of orofacial clefts by Kentucky ADDs. The most noticeable trend is that the rate of clefts is elevated in southeastern Kentucky; a possible contributing factor to this is the high prevalence of smoking. In the three ADDs with the highest rates of smoking (Big Sandy, Kentucky River, and Purchase), the orofacial cleft rate is more than double that of the lowest ADDs (Buffalo Trace and Pennyrile). However, the low rate of Buffalo Trace should be considered an unstable estimate due to the small case numbers and small population in this region.

Map 4: Rate of Orofacial Clefts by Area Development District, Kentucky, 2005-2014

# Rate of Orofacial Clefts by ADD, Kentucky, 2005-2014



February 25, 2016

Data Source: Kentucky Birth Surveillance Registry, Kentucky Live Birth Certificates, Kentucky Stillbirth Certificates

Cases definition: ICD-9 codes 749.00-749.29

Shapefiles from Kentucky Geography Network

Prepared by Emily Ferrell, MPH CPH

Notes: Kentucky residents only. Rates are per 10,000 live births. 2012-2014 data are preliminary and numbers may change.

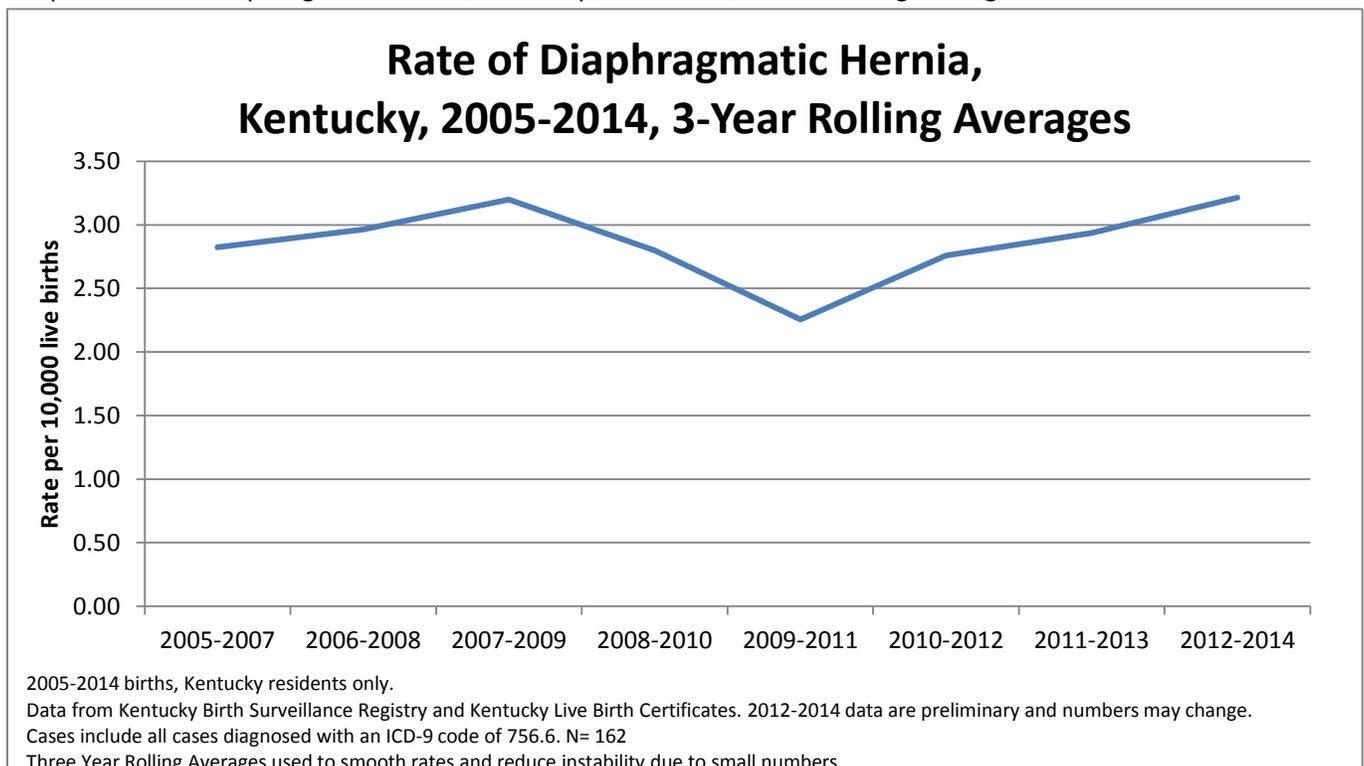
## Diaphragmatic Hernia

The diaphragm is composed of muscle and other fibrous tissue and separates the organs in the abdomen from those in the chest.<sup>34</sup> Abnormal development of the diaphragm before birth can lead to an abnormal opening (hernia) that allows the stomach and intestines to move into the chest cavity and crowd the heart and lungs. One effect of this crowding is underdevelopment of the lungs (pulmonary hypoplasia), which can potentially lead to life-threatening breathing difficulties in newborns.<sup>34</sup> Mortality among live-born infants with diaphragmatic hernia is estimated at 20-50%; additionally, approximately 30% of prenatally-diagnosed cases will result in elective terminations, miscarriages, or stillbirths.<sup>35</sup>

For most individuals with a congenital diaphragmatic hernia, the cause is unknown. In an estimated 10-15% percent of affected individuals, the condition appears as one component of a syndrome.<sup>34</sup> Of those individuals who do not have an underlying syndrome, about 25% have abnormalities of other body systems, including the heart, brain, skeleton, intestines, genitals, kidneys, or eyes.<sup>34</sup>

According to KBSR data from 2005-2014, there are approximately 16 cases a year of diaphragmatic hernia in Kentucky; across the U.S. there are about 1,088 cases a year.<sup>25</sup> The Kentucky and U.S. rates are similar (2.9 and 2.6 cases per 10,000 live births, respectively). Little is known about the risk factors for congenital diaphragmatic hernia. A few studies have indicated an association with advanced maternal age in syndromic cases, and others have indicated that diaphragmatic hernia is more common in male infants.<sup>35</sup> No significant trends have been identified in geographic or racial-ethnic distribution of diaphragmatic hernia cases.<sup>35</sup>

Graph 8: Rate of Diaphragmatic Hernia, Kentucky, 2005-2014, 3-Year Rolling Averages



## Abdominal Wall Defects

An abdominal wall defect is an opening (of any size) in the abdomen through which abdominal organs can protrude.<sup>36</sup> Abdominal wall defects occur early in fetal development and can usually be diagnosed between the tenth and fourteenth weeks of pregnancy.<sup>36</sup> Multiple genetic and environmental factors likely influence the development of abdominal wall defects, although no specific genetic changes are known to be causative.<sup>36</sup> The two most common abdominal wall defects, which will be addressed below, are **gastroschisis** and **omphalocele**.

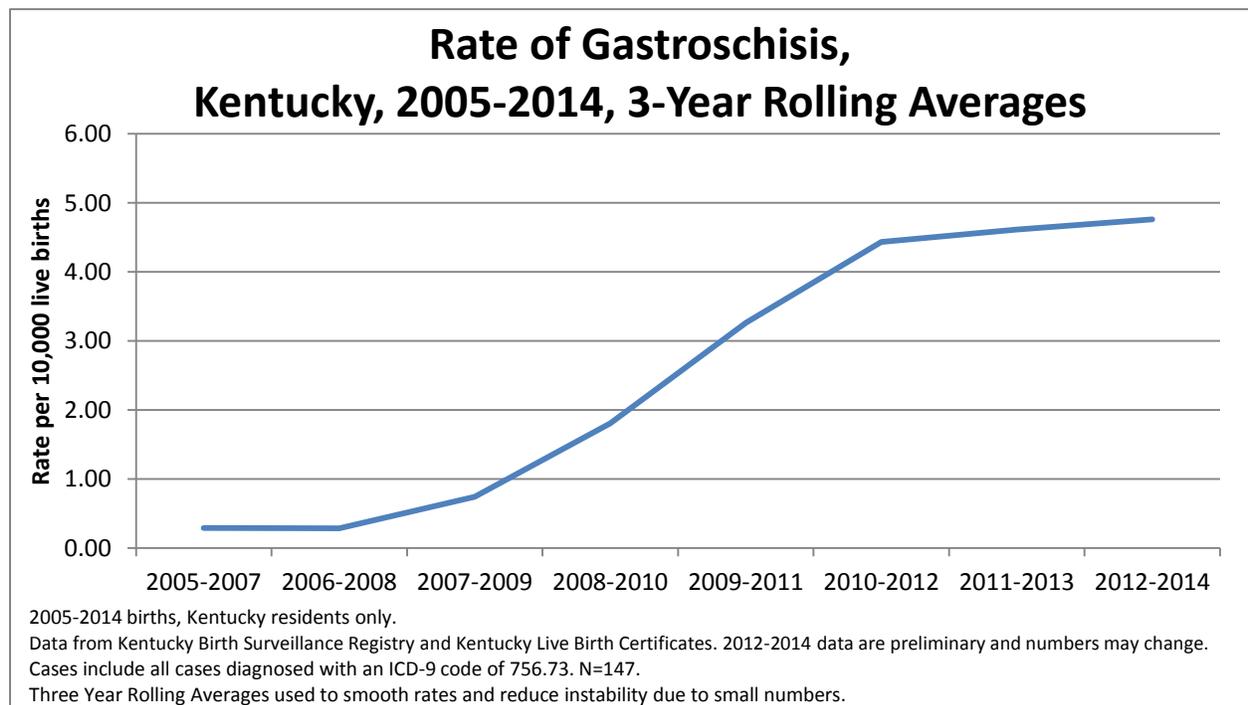
**Gastroschisis:** In this abdominal wall defect, the baby's intestines are outside of the baby's body through a hole beside the belly button.<sup>37</sup> Sometimes other organs, such as the stomach and liver, may be outside of the baby's body. Because the organs are not covered in a protective sac, the bowel can become irritated, causing it to shorten, twist, or swell.<sup>37</sup> Gastroschisis must be surgically repaired soon after birth, which entails placing the abdominal organs inside the body and repairing the abdominal wall.<sup>37</sup> Even after the repair, infants with gastroschisis can have problems with feeding, digestion, and absorption of nutrients.

Figure 4 Gastroschisis



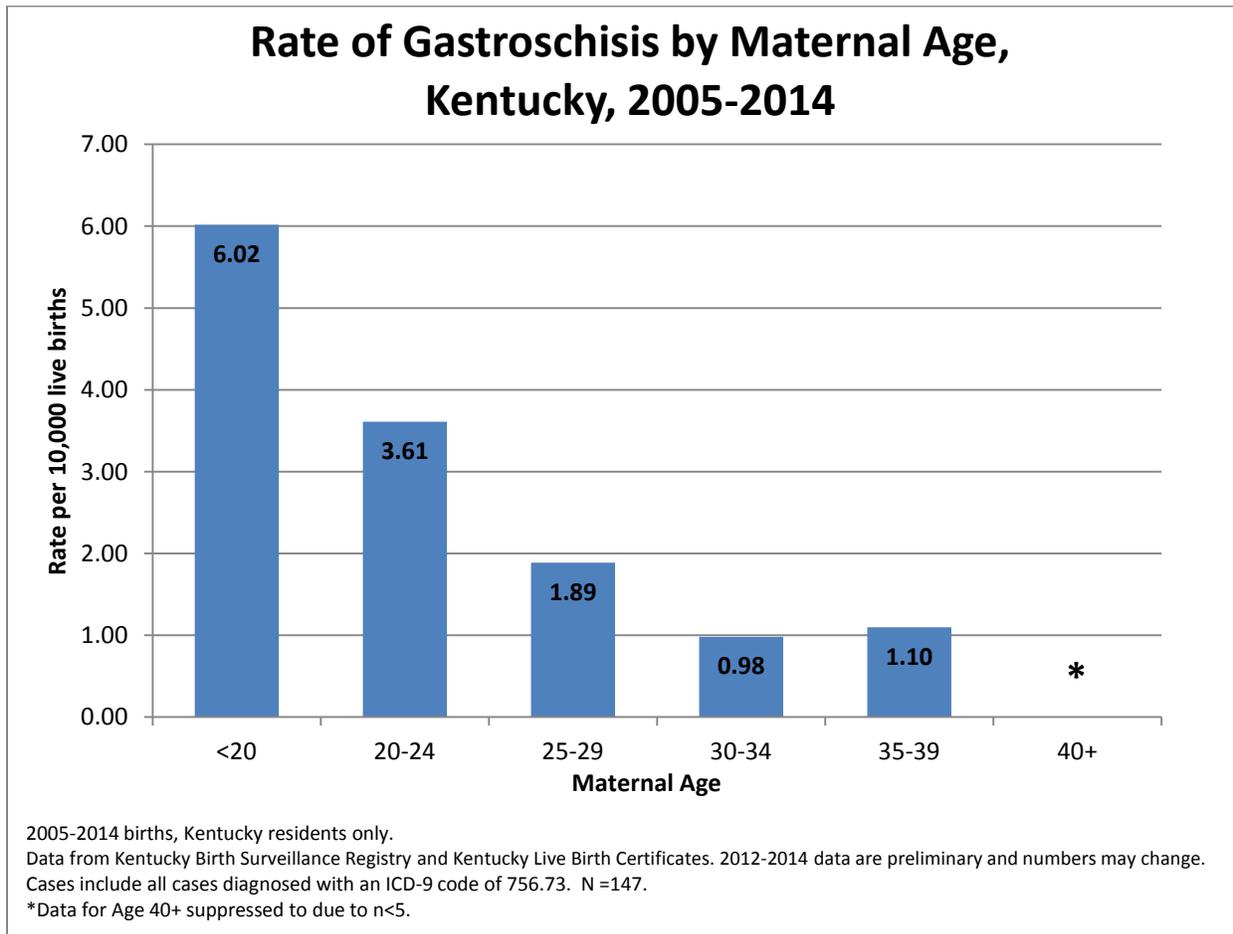
From 2005-2014, there were an average of 15 cases annually in Kentucky; in the U.S., there are approximately 1,871 cases of gastroschisis annually.<sup>25</sup> The corresponding rates are 2.6 and 4.5 cases per 10,000 live births, respectively. According to a study using data across the U.S., from 1995-2012 the prevalence of gastroschisis has been increasing for all maternal age groups but especially young Non-Hispanic Black mothers.<sup>38</sup> KBSR data from 2005-2014 replicates this trend.

Graph 9: Rate of Gastroschisis, Kentucky, 2005-2014, 3-Year Rolling Averages



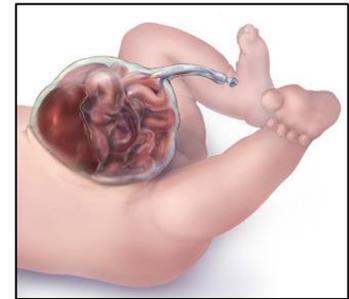
Alcohol and tobacco use during pregnancy are two factors associated with gastroschisis.<sup>37</sup> According to 2005-2014 KBSR data, women who smoke during pregnancy have at least an 80% greater risk of having a child with gastroschisis compared to non-smokers. Another notable risk factor for gastroschisis is young maternal age (<20 years old).<sup>38</sup> In the data below, the rate of gastroschisis in young mothers is approximately six times the rate in mothers over age 30. While mothers age 20-24 also have a higher risk, the difference is not as dramatic.

Graph 10: Rate of Gastroschisis by Maternal Age, Kentucky, 2005-2014



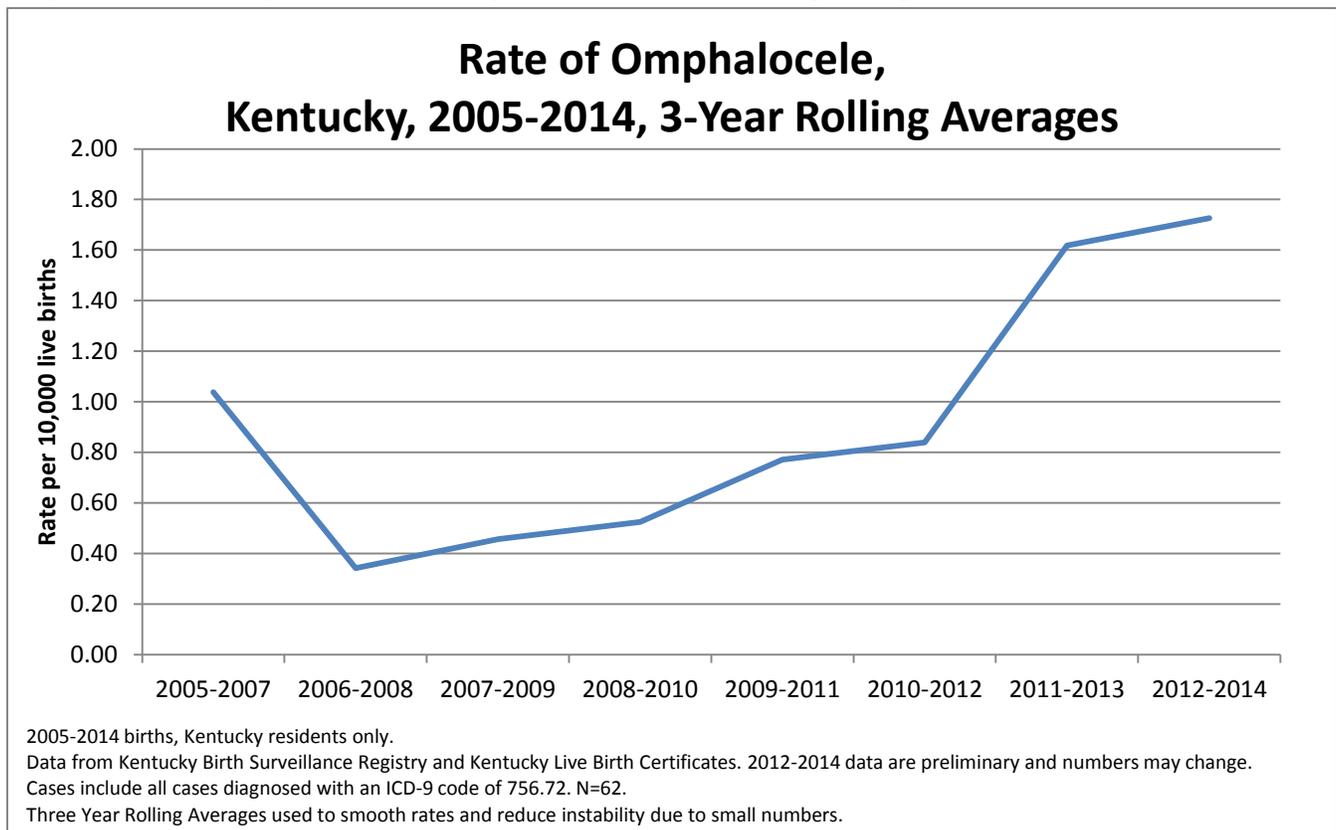
**Omphalocele:** In this abdominal wall defect, the infant’s intestines, liver, or other organs are outside of the belly through the belly button, covered in a thin sac.<sup>39</sup> If the omphalocele is small, it is usually surgically repaired soon after birth, but if the defect is large, the repair may be done in stages.<sup>39</sup> Babies born with an omphalocele can have other related problems such as reduced size of the abdominal cavity, infection, or damaged organs (through pinching or twisting). Omphalocele is often found in children with other defects such as heart defects and neural tube defects.<sup>39</sup> In many cases, omphalocele is one feature in a genetic syndrome.<sup>36</sup>

Figure 5: Omphalocele



From 2005-2014, there were an average of six cases annually in Kentucky; in the U.S., there are approximately 775 cases of omphalocele annually.<sup>25</sup> The corresponding rates are 1.1 and 1.9 cases per 10,000 live births, respectively. The rate of omphalocele in Kentucky has fluctuated from 2005-2014, in part due to the rarity of the condition. Even when calculating 3-year rolling averages, the average number of cases per 3-year period is less than 20 and rates should be considered unstable.

Graph 11: Rate of Omphalocele, Kentucky, 2005-2014, 3-Year Rolling Averages

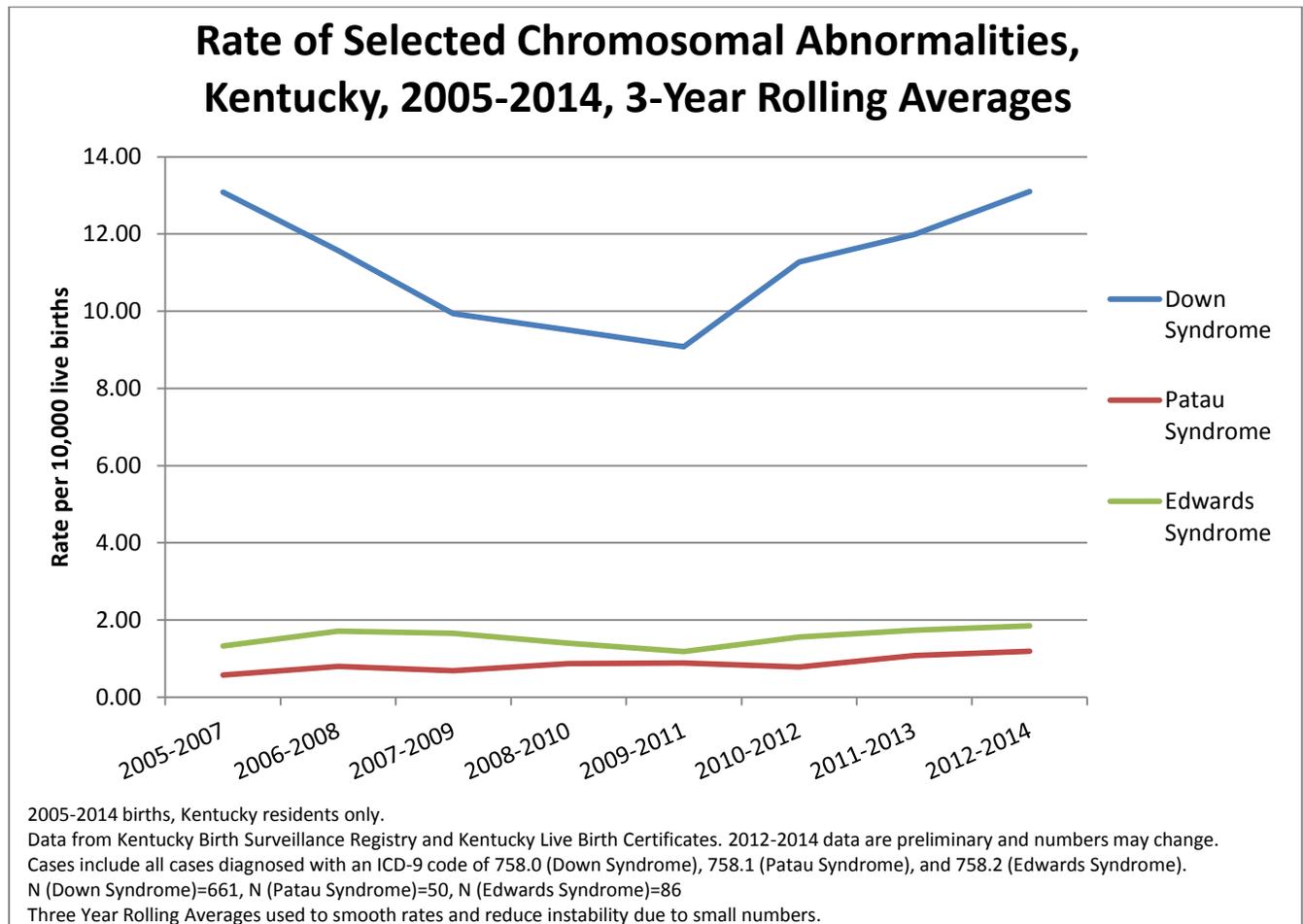


Risk factors for developing omphalocele include maternal alcohol and tobacco use, maternal use of selective serotonin-reuptake inhibitors (SSRIs), and obesity.<sup>39</sup> Kentucky data from 2005-2014 indicates that women who smoked during pregnancy had 71% increased risk of delivering a baby with an omphalocele.

## Chromosomal Abnormalities

Chromosomes are the structures that carry all genetic information for an individual. An individual's chromosomes are in every cell of the body and instruct the body on growth and development. The typical individual has a total of 46 chromosomes (23 pairs of chromosomes). Individuals inherit one copy of each chromosome pair from their mother and the other copy from their father. Sometimes in development, an error occurs, and a baby has too much or too little chromosomal material. When an individual has three copies instead of two for a particular chromosome, it is called a trisomy. Trisomy conditions can affect the normal growth of the body and brain, causing mental and physical challenges.<sup>40</sup> **Down Syndrome (Trisomy 21), Patau Syndrome (Trisomy 13), and Edwards Syndrome (Trisomy 18)** are the three most common trisomy conditions<sup>40</sup> and are explained in more detail below.

Graph 12: Rate of Selected Chromosomal Abnormalities, Kentucky, 2005-2014, 3-Year Rolling Averages



**Down Syndrome (Trisomy 21):** The most common trisomy disorder is Down Syndrome. While it affects people differently, most individuals have characteristic physical features and some degree of developmental delay. Common physical features include: a flattened face, almond-shaped eyes that slant up, a short neck, small ears, a tongue that tends to stick out of the mouth, tiny white spots on the eye's iris, small hands and feet, a single line across the palm of the hand, poor muscle tone or loose joints (hypotonia), and shorter height.<sup>40</sup> Babies born with Down syndrome have a higher risk for heart defects, stomach and digestive problems, eye problems, and hypothyroidism.<sup>41</sup> Ear infections and hearing loss are also common.<sup>40</sup>

***Public Health Feature: Information for Families***

In June 2013, legislation was passed requiring that expectant and new parents of children with Down Syndrome receive information about educational and support services in Kentucky.<sup>42</sup>

KRS 211.192 states that “any health facility as defined in KRS 216B.015(13), physician, health care provider, nurse midwife, or genetic counselor who renders prenatal care, postnatal care, or genetic counseling, upon receipt of a positive test result from a test for Down syndrome, shall provide the expectant or new parent with information provided by the Cabinet for Health and Family Services.”<sup>42</sup>

This information was compiled to educate families about Down Syndrome, provide sources for further information, and provide contact information for community resources that could provide familial support.

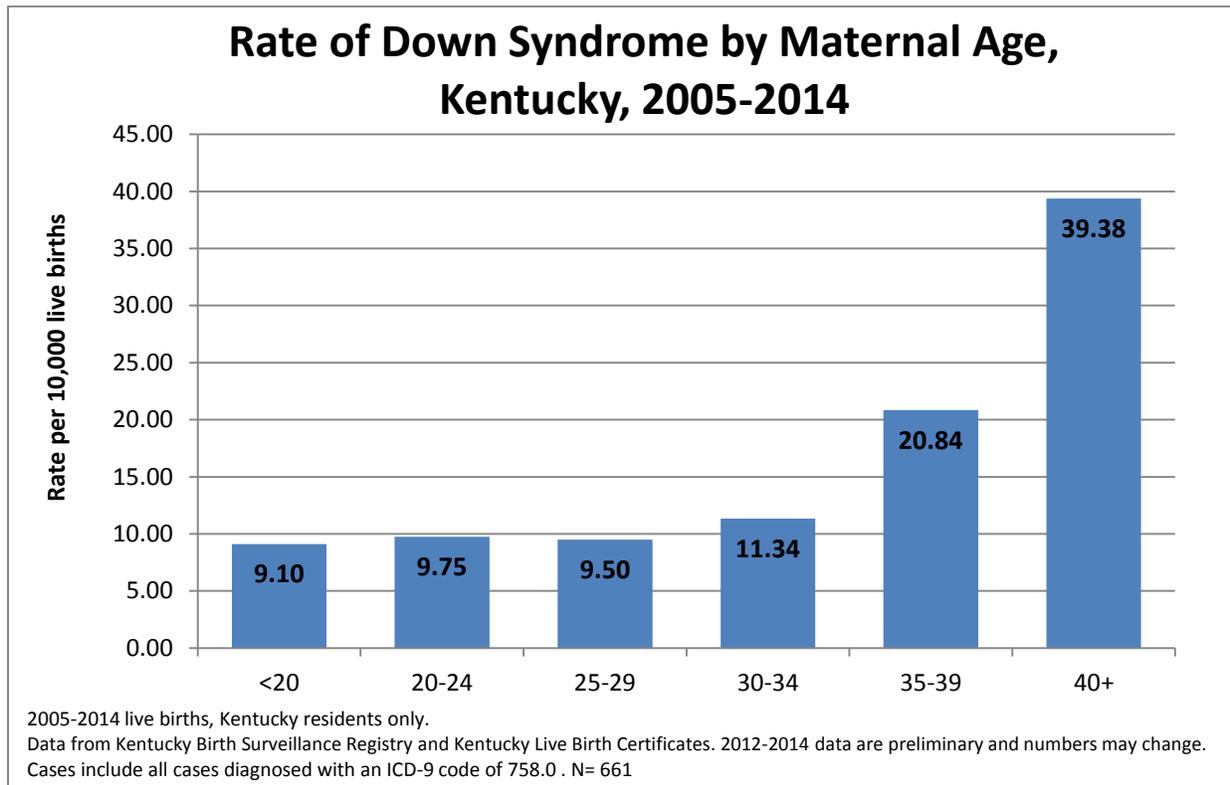
This information can be found at the following website:

<http://chfs.ky.gov/dph/mch/e cd/DownSyndromeInformation.htm>

About 66 children are born with Down Syndrome each year in Kentucky or about 11.6 cases per 10,000 live births. Across the U.S., 6,037 children are born with Down Syndrome each year (about 14.5 cases per 10,000 live births).<sup>25</sup>

The risk of having a child with Down Syndrome increases with maternal age. However, most babies with Down Syndrome have mothers who are younger than 35, because there are many more births among younger women.<sup>40</sup> According to KBSR data from 2005-2014, mothers in the 35-39 year old age group have more than twice the risk of having a child with Down Syndrome versus mothers who are 25-29. Among mothers who are 40 years old or greater, the risk is magnified by four times compared to 25-29 year olds.

Graph 13: Rate of Down Syndrome by Maternal Age, Kentucky, 2005-2014



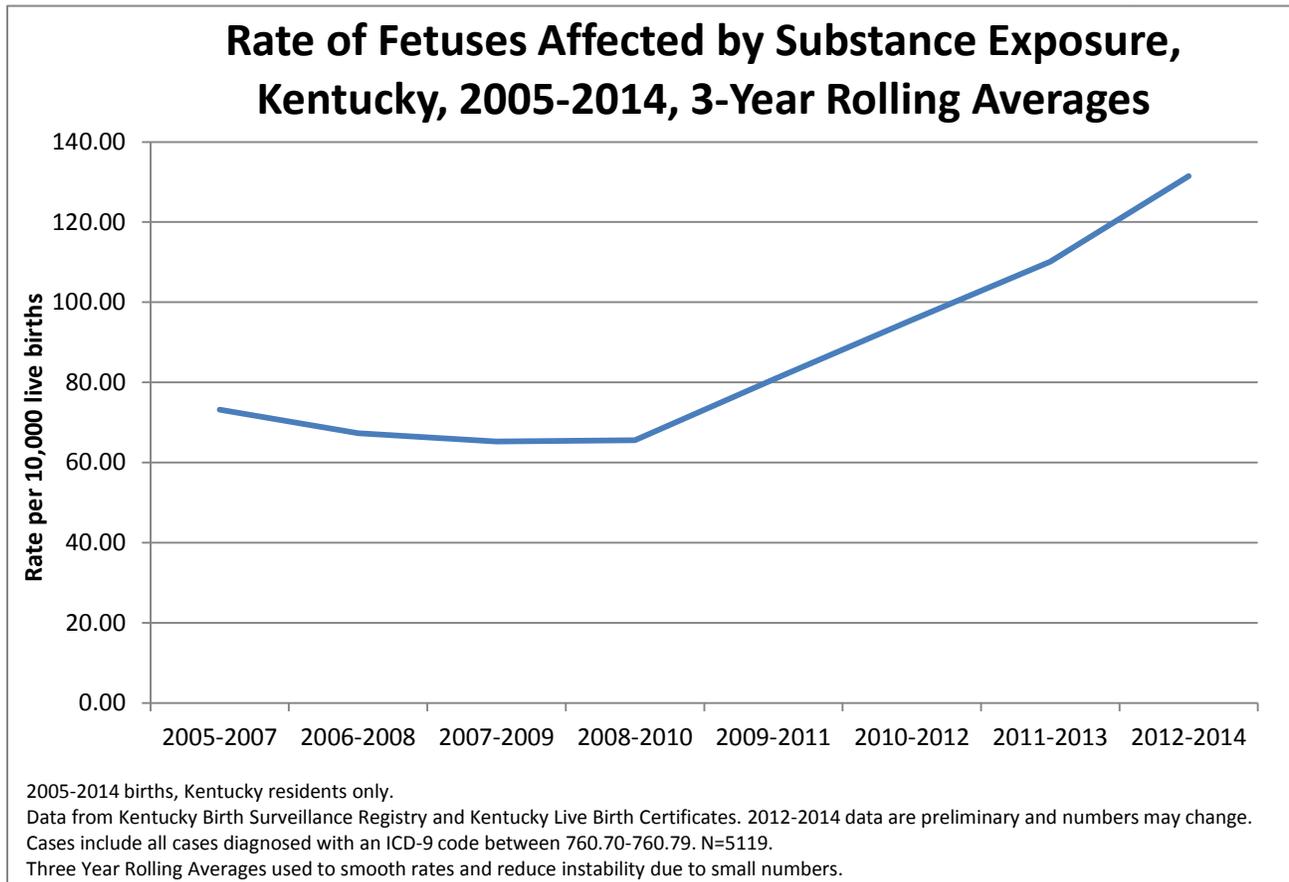
**Patau Syndrome (Trisomy 13):** This is a chromosomal condition associated with severe intellectual disability and physical abnormalities affecting multiple body systems.<sup>43</sup> Individuals often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes (polydactyly), orofacial clefts, and hypotonia.<sup>43</sup> Due to the association with life-threatening medical problems, most infants with Patau Syndrome die within days or weeks of birth; only 5-10% of cases survive beyond their first birthday.<sup>43</sup> Patau Syndrome is very rare; about five children are born with it each year in Kentucky (528 in the U.S.).<sup>25</sup> In both Kentucky and the U.S., only about 1 case per 10,000 live births has the condition.

**Edwards Syndrome (Trisomy 18):** This is a chromosomal condition associated with severe intellectual disability and abnormalities in many body systems.<sup>44</sup> Individuals with Edwards Syndrome often have slow growth before birth, low birth weight, heart defects, and other congenital anomalies.<sup>44</sup> Due to the presence of several life-threatening medical problems, many cases are stillborn or die within the first month of life. Only 5-10% of children with this condition survive infancy.<sup>44</sup> Edwards Syndrome is a rare condition; there are about 9 children born with it annually in Kentucky and about 1,109 are born each year in the U.S.. The rate of Edwards Syndrome is 1.5 cases per 10,000 live births in Kentucky versus 2.7 cases per 10,000 live births in the U.S.<sup>25</sup>

## Fetuses Affected by Substance Exposure

Substance abuse has reached epidemic levels in Kentucky and across the nation. In the 2015 Title V Needs Assessment for communities across the state, substance abuse was the top concern over all other health issues among all maternal and child health populations. Using KBSR data, it is estimated that each year in Kentucky over 10,000 infants are exposed to substances (including tobacco) prenatally; due to underreporting, this number could actually be much greater. The graph below excludes children only exposed to tobacco prenatally but includes children affected by alcohol, narcotics, hallucinogenic agents, anti-infectives, cocaine, diethylstilbestrol, anticonvulsants, antimetabolic agents, and other noxious influences (defined by ICD-9 codes 760.70- 760.79). The rate of fetuses affected by substance exposure has been increasing from 2005-2014. This trend is concerning as the relationship between fetal substance exposure and birth defects is not well understood.

Graph 14: Rate of Fetuses Affected by Substance Exposure, Kentucky, 2005-2014, 3-Year Rolling Averages



The three types of substance exposures that will be addressed below include **alcohol, medication, and tobacco.**

**Alcohol Exposure:** Fetal alcohol spectrum disorders (FASDs) are a group of conditions that may occur in people whose mothers drank alcohol during pregnancy.<sup>45</sup> Physical symptoms include: abnormal facial features including smooth philtrum (area between nose and upper lip); small head size; shorter-than-average height; low body weight; vision or hearing problems; and problems with the heart, kidneys, or bones.<sup>45</sup> Individuals with FASDs might also exhibit problems with coordination, reasoning and judgment skills, and memory.<sup>45</sup> They might also have hyperactive behavior, difficulty paying attention, learning disabilities, speech and language delays, and intellectual disability.<sup>45</sup> The age at diagnosis can vary based on the severity of the child's symptoms, but children are often not diagnosed at birth.<sup>45</sup> Frequently, children are not diagnosed until they miss developmental milestones or exhibit difficulties at school.

Because symptoms or delays are often documented at office visits, hospital discharge data does not provide an accurate estimate of FASD prevalence. Therefore, it is unknown exactly how many people have FASDs. In certain areas of the U.S., CDC studies have identified the rate of Fetal Alcohol Syndrome (FAS), which is the most severe of the FASDs, as 0.2-1.5 cases per 1,000 live births.<sup>46</sup> Estimates for the prevalence of all FASDs range from 6 to 9 cases per 1,000 live births.<sup>46</sup>

Data from the Behavioral Risk Factor Surveillance System (BRFSS) indicates that 1 in 10 pregnant women consumes alcohol during pregnancy in the U.S..<sup>47</sup> Due to small sample sizes, Kentucky-specific BRFSS data is not available as a comparison. Data from the a 2013 study by the Substance Abuse and Mental Health Services Administration (SAMHSA) has findings similar to the BRFSS: on average in the U.S., 9.4% of pregnant women reported current alcohol use, 2.3% reported binge drinking, and 0.4% reported heavy drinking.<sup>48</sup> These rates were lower than the rates for non-pregnant women in the same age group (55.4%, 24.6%, and 5.3%, respectively). About half of all U.S. pregnancies are unplanned so most women do not know they are pregnant until they are 4-6 weeks into the pregnancy.<sup>49</sup> Even among planned pregnancies, women usually do not know they are pregnant for several weeks and might be drinking and exposing the developing baby to alcohol without knowing it. The SAMSHA study supports this conclusion as it finds that alcohol use was lower among pregnant women during the second and third trimesters than during the first trimester (5.0% and 4.4% vs. 19.0%, respectively).<sup>48</sup>

The lifetime cost for one individual with FAS has been estimated to be \$2 million; it is estimated that the U.S. cost for FAS alone is over \$5.5 billion annually.<sup>49</sup>

**Medication Exposure:** Fewer than 10% of medications approved by the U.S. Food and Drug Administration (FDA) have enough information to determine their safety for use in pregnancy.<sup>50</sup> However, about 90% of pregnant women take at least one medication or supplement of any kind, and about 70% of pregnant women take at least one prescription medicine, a percent which has been increasing in recent decades.<sup>50</sup> Annually in the U.S., 5.4 million pregnancies are exposed to medication.<sup>50</sup>

Not all medication use during pregnancy is harmful; many women need medication to appropriately manage a health condition. Women should talk to their health care provider to determine what is best

for their situation.<sup>4</sup> In some cases, stopping medication use during pregnancy may be more harmful than taking the medication.<sup>4</sup> However, use of certain medications during pregnancy can cause serious birth defects or poor pregnancy outcomes.

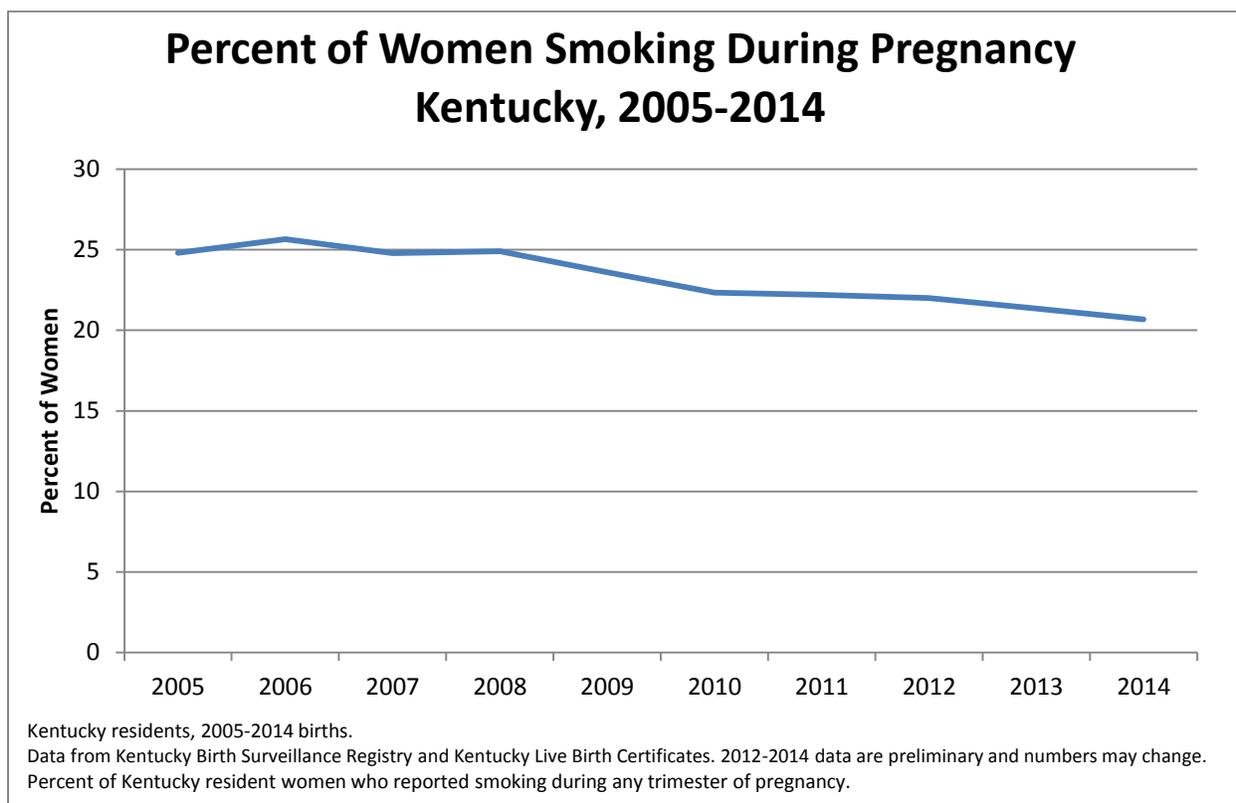
Medications that are possibly associated with birth defects include:

- Selective serotonin-reuptake inhibitors (antidepressants)— linked to an increased risk of heart defects, anencephaly, and abdominal wall defects.<sup>51</sup>
- Corticosteroids (prescribed for asthma)—might increase the risk of orofacial clefts.<sup>51</sup>
- Antiepileptic or anticonvulsant medications such as valproic acid and carbamazepine—increased risk for multiple birth defects including NTDs, heart defects, and cleft lip.<sup>51</sup>
- Progestins (infertility treatments and birth control)—possible increased risk for hypospadias.<sup>51</sup>
- Isotretinoin (brand name Accutane®)—causes birth defects in more than 35% of infants whose mothers take the drug during pregnancy, including small or absent ears (microtia or anotia), a small jaw (micrognathia), small head (microcephaly), and cleft palate.<sup>52</sup>
- Methotrexate (immunosuppressant)— associated with malformations of the head, face, and bones; decreases the body's ability to break down and use folic acid.<sup>53</sup>
- Thalidomide (brand name Thalamid®)—there is a risk of 20% or greater to have a baby with birth defects such as extremely short or missing arms and legs, anotia, heart defects, missing or small eyes (anophthalmia or microphthalmia), and kidney abnormalities.<sup>54</sup>

**Tobacco Exposure:** Tobacco exposure is by far the most common harmful prenatal substance exposure in the U.S. and especially in Kentucky. Smoking reduces the amount of oxygen available to the mother, and therefore the fetus. It is associated with negative pregnancy outcomes such as low birth weight, prematurity, placenta previa, and placental abruptions<sup>15</sup> and increases the risk of having a baby with certain birth defects, including heart defects, orofacial clefts, and gastroschisis.<sup>14, 28, 37</sup>

Data from the 2014 Live Birth Certificates indicates that in the U.S., 8.4% of women smoked at any time during pregnancy.<sup>55</sup> Kentucky traditionally has much higher rates of smoking during pregnancy than the rest of the nation, and more than 1 in every 5 Kentucky infants (20.7%) are exposed to cigarette smoke in utero. The percent of women who quit smoking during pregnancy is much lower in Kentucky (11.4%) than in the U.S. as a whole (20.6%).<sup>55</sup> As shown in the graph below, the percent of Kentucky resident women smoking during pregnancy has decreased from about 25% in 2005 to about 20% in 2014.

Graph 15: Percent of Mothers Smoking During Pregnancy, Kentucky, 2005-2014



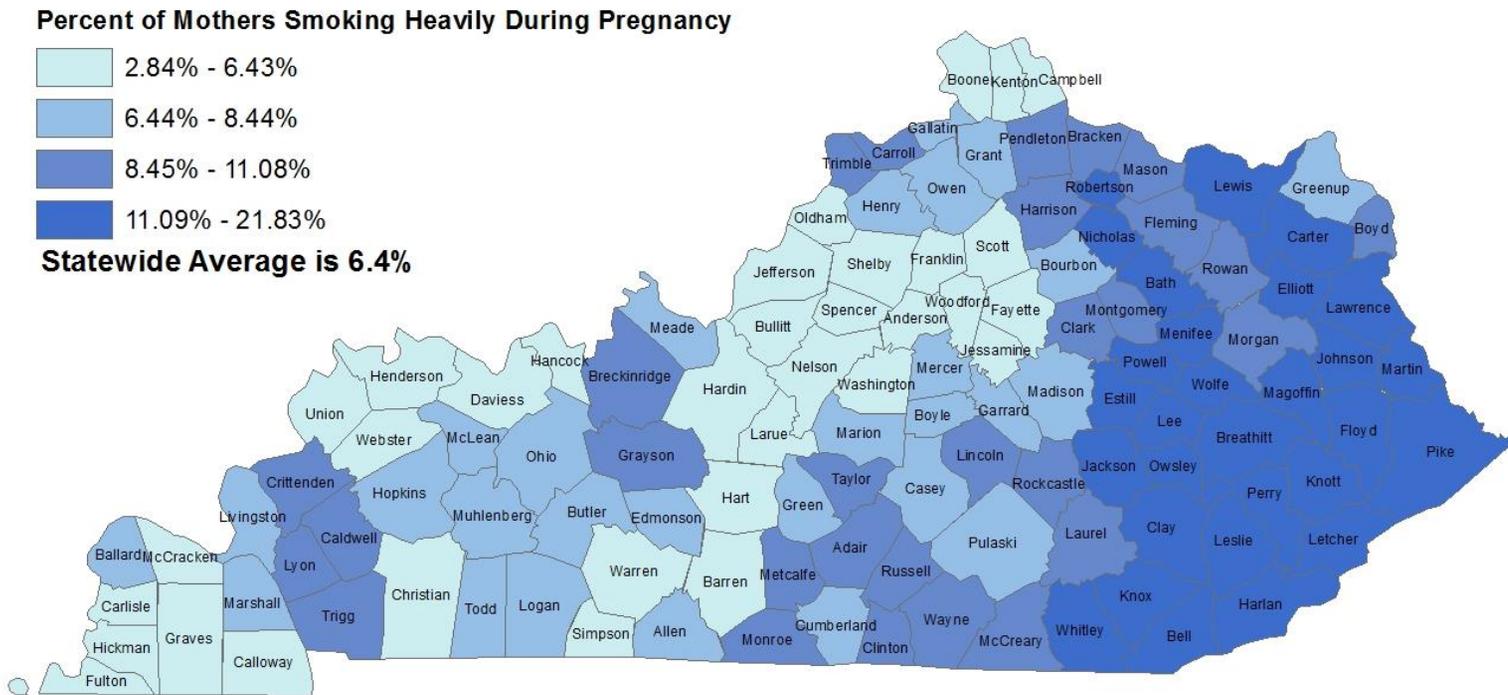
Map 6 shows the percent of mothers per county that smoke during pregnancy using 2014 Kentucky live birth certificate data. In this map, the highest rates of smoking during pregnancy are in eastern Kentucky and the lowest rates are in central and western Kentucky. There is a noticeably large difference in rates across counties shown on this map (values range from 9.4% to 44.1%). This difference may be partially due to comprehensive smoke-free ordinances passed by counties and cities throughout Kentucky. The state average is 20.7%, and only 29 of the 120 counties are below that rate (shown in the lightest blue shade). Jefferson and Fayette, the two most populous counties in Kentucky, help lower the statewide average as their rates are 12.0% and 11.5%, respectively.

Map 7 shows the percent of mothers in each county that are considered heavy smokers (smoking >10 cigarettes per day during the last trimester of pregnancy) using data from 2005-2014 Kentucky live birth certificates. As in the map of smoking rates during pregnancy, the highest rates are in eastern Kentucky and the lowest rates are in central and western Kentucky, although the trend is even more noticeable in this map than on the previous one. The statewide average percent of women who are heavy smokers during pregnancy is 6.4%. Of the 120 Kentucky counties, 34 are below that average (shown in the lightest blue shade). The highest percent of heavy smokers in any county is 21.8%—that equates to more than 1 in every 5 pregnant women smoking heavily during pregnancy.



Map 7: Heavy Smoking During Pregnancy by Mother's County of Residence, Kentucky, 2005-2014

# Heavy Smoking During Pregnancy by Mother's County of Residence; Kentucky, 2005-2014



February 29, 2016

Data Source: Kentucky Live Birth Certificates, 2005-2014. Note: 2012-2014 data are preliminary and numbers may change.  
Percent of Kentucky resident women who reported smoking >10 cigarettes per day during the last trimester of pregnancy.  
Shapefiles from Kentucky Geography Network  
Prepared by Emily Ferrell, MPH CPH

## Partners and Resources:

Kentucky is fortunate to have qualified, capable, and compassionate citizens and professionals who work to ensure that children with birth defects get the care needed for them to thrive. Contact one or more of these groups for more information about how they serve families in Kentucky.



**First Steps** is a statewide early intervention system that provides services to families and children with developmental disabilities from birth to age 3. First Steps serves approximately 7,000 children in Kentucky each year.<sup>56</sup> This program provides screening, referrals, and services to children who have a delay in communication, cognition, physical, social and emotional, or self-help; children are also eligible if they have been diagnosed with a condition that has a high probability of resulting in delays. **KBSR** works with First Steps to identify and refer children who might be eligible for services. Please visit the First Steps website for more information (<http://chfs.ky.gov/dph/firstSteps/default.htm>).



The **Kentucky Commission for Children with Special Health Care Needs (CCSHCN)** provides specialty care to children with a variety of physical disabilities. They offer specialty clinics in 12 regional offices across the state. Clinics are staffed by multi-disciplinary teams to provide comprehensive, coordinated care for a variety of conditions. CCSHCN also provides augmentative therapies and transition support for the young person preparing for adulthood. A parent, physician, or other caregiver can make referrals. Please visit the CCSHCN website for more information (<http://chfs.ky.gov/ccshcn/>).



The **Kentucky Folic Acid and Perinatal Partnership (KFAP)** provides leadership and action for the statewide folic acid campaign in order to prevent neural tube defects and premature birth and promote perinatal health. Please visit the KFAP website for more information (<http://kfap.org/>).

**Kentucky Newborn Screening Program** helps parents identify if a baby has certain health problems. A healthy-looking newborn can have a serious disease that may not be detected without specific screening. If left undetected and untreated, these diseases can lead to slow growth, blindness, brain damage, or possibly death. Early treatment can help prevent these serious problems. Please visit the newborn screening program website for more information (<http://chfs.ky.gov/dph/mch/ececd/newbornscreening.htm>).



The **Kentucky Fetal Alcohol Spectrum Disorders (KYFASD) Center** is responsible for providing training, resources, and technical assistance to agencies and programs statewide. Their core activity

is connecting people with information that will bring light to the issue of fetal alcohol spectrum disorders (FASDs). They provide support for families dealing with FASDs and encourage prevention, advocacy, and awareness. Please visit the KYFASD Center website for more information (<http://www.kyfasd.org/>)

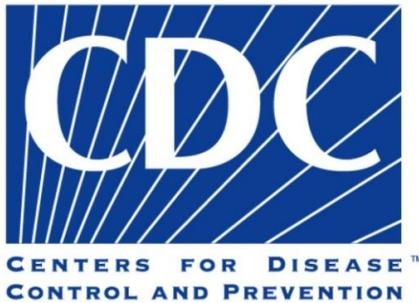


The mission of the **March of Dimes** is to improve the health of babies by preventing birth defects, premature birth, and infant mortality. In the field of birth defects, the March of Dimes funds research, promotes newborn screening, and educates medical

professionals and the public about best practices for a healthy pregnancy. Please visit the March of Dimes, Kentucky Chapter's website for more information (<http://www.marchofdimes.org/kentucky/>).



The **National Birth Defects Prevention Network (NBDPN)** maintains a national network of state and population-based programs for birth defects surveillance and research. NBDPN assesses the impact of birth defects on children, families, and health care; identifies factors that can be used to develop primary prevention strategies; and assists families and their providers in secondary disabilities prevention. Please visit the NBDPN website for more information (<http://www.nbdpn.org>).



The **Centers for Disease Control and Prevention (CDC)** and the National Center on Birth Defects and Developmental Disabilities monitor and research birth defects in the U.S.. **KBSR** is one of 14 states that currently receives CDC funding to assist in the implementation of a birth defects registry. KBSR sends accurate and high-quality data to the CDC, which can be used in nationally-representative studies. Please visit the CDC's National Center on Birth Defects and Developmental Disabilities website for more information (<http://www.cdc.gov/ncbddd>).

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## The Cost of Birth Defects

Birth defects cause pain and suffering for families, and the financial burden of caring for children with birth defects can be substantial. Often, babies born with birth defects need special treatments or services to thrive, adding to the costs of their care. The costs might be for a one-time surgery, or they might continue across a lifetime as ongoing treatment and intervention is needed. Families, communities, and the government share these costs; prevention of birth defects is critical.

In the U.S., birth defects led to more than 139,000 hospital stays during a single year (based on 2004 data), resulting in \$2.6 billion in hospital costs alone.<sup>57</sup> According to a more recent study using Texas data, over a 10-year period hospital charges for children with birth defects totaled \$24.8 billion.<sup>58</sup> Overall, the mean of total hospital charges was approximately six times greater for children with birth defects than for children without birth defects.<sup>58</sup> One component of the elevated costs is increased length of stay, as mean hospital stay for children with birth defects was more than twice that of those without.<sup>58</sup>

Examples of costs of birth defects are below:

Heart defects:

- A study using national data showed that overall hospital costs for all people with a congenital heart defect in the U.S. were about \$1.4 billion in a single year.<sup>57</sup>

Spina bifida:

- A study using Florida data showed average hospital costs in the first year of life for a baby born with spina bifida were about \$21,900, but costs ranged up to over \$1.3 million.<sup>59</sup>
- The total lifetime cost of care for a child born with spina bifida is estimated to be \$706,000, according to a study using South Carolina data.<sup>60</sup>

Down syndrome:

- In a study using national U.S. data, the medical costs for a child with Down syndrome have been estimated as 12-13 times greater than for a child without Down syndrome.<sup>61</sup>

Multiple defects:

- According to a study in Florida, children with Down syndrome and a severe CHD had 11 times higher hospital costs during the first year of life when compared to children with isolated Down Syndrome.<sup>62</sup>
- In the same study, children with Down syndrome and a major birth defect other than a CHD had 4 times higher hospital costs during the first year of life when compared to children with isolated Down Syndrome.<sup>62</sup>

These figures cannot estimate the emotional cost to the children with birth defects and their families, but they help illustrate the magnitude of the problem and the need for prevention. Preventing birth defects is good for babies. It's good for families. It's good for our communities.

## Future Directions for KBSR:

The Kentucky Birth Surveillance Registry has served as the primary source of birth defects data in the Commonwealth of Kentucky for nearly 20 years. Through surveillance, referrals, prevention, and research, KBSR works to expand Kentuckians' understandings of the trends and risk factors of birth defects.

KBSR strives to improve data quality and timely surveillance measures by focusing on strategies that will reduce time to case ascertainment, increase case completeness, and ensure accuracy in recorded diagnoses. Specific activities planned for the future include: partnering with hospitals in neighboring states to improve reporting for Kentucky residents who seek out-of-state medical care, improving linkage of KBSR cases to birth and death certificates, identifying and researching cases of FASDs and other fetal substance exposure disorders, expanding the number of conditions abstracted and improving data abstraction methodologies, expanding the number of cases identified for referrals to services by working with additional state agencies, exploring the possibility of active surveillance, and developing targeted prevention activities for specific populations who are at increased risk for birth defects.

Two particular areas of focus in the future for KBSR are **Neonatal Abstinence Syndrome (NAS)** and **Microcephaly/Zika virus**, both described below. These two topics are emerging public health issues that will need multidisciplinary cooperation to research, understand, and prevent. KBSR staff members are involved in projects regarding both of these topics, and KBSR data can be used to enhance the understanding of the topics and to complement data from other agencies. As a registry that tracks children through age five, KBSR is often the only data source that has long-term follow-up information on the cases.

**Neonatal Abstinence Syndrome (NAS):** NAS is a collection of symptoms babies experience in withdrawing from drugs they were chronically exposed to in utero.<sup>63</sup> When the umbilical cord is cut at delivery, these substances no longer circulate to the baby, and the baby has to re-adjust to a drug-free environment. Symptoms can include a high-pitched cry, restlessness, hyperactive reflexes, myoclonic jerks, jitteriness, tremors, seizure, poor feeding, vomiting, loose stools, fever, sweating, mottling, nasal flaring, apnea, and tachypnea.<sup>63</sup>

According to the *Kentucky Annual Report From the Public Health NAS Reporting Registry*, there were 1,234 unduplicated cases of NAS reported from July 2014 to June 2015.<sup>64</sup> These estimates are very conservative as under-reporting is a concern. The large number of cases has led to NAS becoming an area of public health interest, and Kentucky has enacted several laws in order to better understand and combat the growing epidemic of substance abuse.

### ***Public Health Feature: NAS Surveillance***

As of July 2014 in Kentucky, NAS is a public health reportable disease/condition (KRS 211.676).<sup>65</sup> That means that data can be collected by the Department for Public Health to understand the extent of the problem. Another law, KRS 211.678, calls for an annual report of de-identified, aggregate statistical data from this reporting.<sup>66</sup> This data report can inform policymakers about how to better address the issue of NAS in Kentucky.

There is little research on associations between NAS and birth defects, but some specific prenatal drug exposures have been associated with birth defects.<sup>63</sup> As KBSR collects information on cases affected by prenatal substance exposure, the database has information about potential NAS cases. Data from KBSR can be compared to data from the NAS surveillance system to identify under-reporting in either database. Chart abstraction on KBSR cases will also provide a method for KBSR to ensure that diagnosis codes are being assigned accurately and consistently. For example, cases diagnosed with NAS should be checked to see if they have also been diagnosed as affected by exposure to narcotics, an unspecified substance, or another substance (ICD-9 codes 760.70, 760.72, or 760.79).

**Microcephaly and Zika virus:** Microcephaly is the clinical finding of a small head compared with infants of the same sex and gestational age. Obtaining a measurement of an infant's head circumference is considered a reliable assessment of the volume of the underlying brain.<sup>67</sup> Infants with severe microcephaly have brains that did not develop properly during pregnancy. Depending on the severity of the microcephaly, these babies can experience a range of health problems including seizures, developmental delay, intellectual disability, problems with movement and balance, feeding problems, hearing loss, and vision problems.<sup>68</sup>

Recently there has been a widespread epidemic of Zika virus reported in South America, Central America, the Caribbean, and the Pacific islands.<sup>69</sup> A major concern associated with this virus is a notable increase in microcephaly among the infants born to mothers infected with Zika. The CDC has recently announced that there is a causal relationship between Zika and microcephaly.<sup>70</sup> The Zika virus is primarily spread through the bite of an infected *Aedes* mosquito. Symptoms of Zika among adults include fever, rash, joint pain, and conjunctivitis (red eyes).<sup>69</sup> These symptoms are so mild and similar to symptoms of many other infections many cases may not be recognized or reported.

In response to this outbreak, the CDC is developing a national Pregnancy and Zika Virus Disease Surveillance Registry to closely monitor Zika among pregnant women in the U.S. and track birthing outcomes. In addition, KBSR is working with the CDC to better monitor infants born with microcephaly in Kentucky. KBSR is exploring ways to more actively track congenital microcephaly cases to better identify risk factors and outcomes. KBSR staff will be involved in long-term follow-up and monitoring of infants born to mothers who are Zika-positive or Zika-inconclusive.

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## Appendix 1: Regulations- Kentucky Revised Statutes 211.651-670

### 211.651 Definitions for KRS 211.651 to 211.670.

As used in KRS 211.651 to 211.670, unless the context otherwise requires:

- (1) "Cabinet" means the Cabinet for Health and Family Services;
- (2) "Secretary" means the secretary of the Cabinet for Health and Family Services;
- (3) "Department" means the Department for Public Health; and
- (4) "Designee" means a local health department, board for mental health or individuals with an intellectual disability, or other governmental or private agency designated by the Department for Public Health.

**Effective:** July 12, 2012

**History:** Amended 2012 Ky. Acts ch. 146, sec. 100, effective July 12, 2012. -- Amended 2005 Ky. Acts ch. 99, sec. 367, effective June 20, 2005. -- Amended 1998 Ky. Acts ch. 426, sec. 309, effective July 15, 1998. -- Created 1992 Ky. Acts ch. 62, sec. 1, effective July 14, 1992.

### 211.655 Legislative findings and statement of intent.

The General Assembly hereby finds and declares that congenital anomalies, stillbirths, and high risk conditions represent problems of significant health importance about which too little is known; that conditions of this nature lead to severe mental anguish on the part of parents and frequently to high medical care costs; and that a system to obtain more information about these conditions could result in development and implementation of preventive measures to decrease their incidence and prevalence in the future. Therefore, it is the intent of the General Assembly to:

- (1) Provide information on the incidence, prevalence, and trends of birth defects, stillbirths, and high risk conditions;
- (2) Provide information as to the possible causes of congenital anomalies, stillbirths, and high risk conditions; and
- (3) Develop prevention strategies to reduce the incidence of congenital anomalies, stillbirths, and high risk conditions and to reduce secondary complications associated with high risk conditions.

**Effective:** July 15, 1994

**History:** Amended 1994 Ky. Acts ch. 405, sec. 76, effective July 15, 1994. -- Created 1992 Ky. Acts ch. 62, sec. 2, effective July 14, 1992.

**211.660 Kentucky birth surveillance registry -- Department's authority to promulgate administrative regulations.**

- (1) The Department for Public Health shall establish and maintain a Kentucky birth surveillance registry that will provide a system for the collection of information concerning birth defects, stillbirths, and high-risk conditions. The system may cover all or part of the Commonwealth.
- (2) In establishing the system, the department may review vital statistics records, and shall also consider expanding the current list of congenital anomalies and high-risk conditions as reported on birth certificates.
- (3)
  - (a) The department may require general acute-care hospitals licensed under KRS Chapter 216B to maintain a list of all inpatients and voluntarily to maintain a list of all outpatients up to the age of five (5) years with a primary diagnosis of a congenital anomaly or high-risk condition as defined by the department upon the recommendation of the appointed advisory committee. Hospital participation regarding its outpatients shall be voluntary and subject to the discretion of each hospital.
  - (b) The department may require medical laboratories licensed under KRS Chapter 333 to maintain medical records for all persons up to the age of five (5) years with a primary diagnosis of or a laboratory test result indicating congenital anomaly or high-risk condition as defined by the department upon the recommendation of the appointed advisory committee.
- (4) Each licensed free-standing birthing center, general acute-care hospital licensed under KRS Chapter 216B, and medical laboratory licensed under KRS Chapter 333 shall grant, if required or otherwise participating voluntarily under the provisions of subsection (3) of this section, to any Kentucky Birth Surveillance Registry personnel or his or her designee, upon presentation of proper identification, access to the medical records of any patient meeting the criteria in subsection (3) of this section. If the department's agent determines that copying of the medical records is necessary, associated costs shall be borne by the Department for Public Health at the rate pursuant to KRS 422.317.
- (5) No liability of any kind, character, damages, or other relief shall arise or be enforced against any licensed free-standing birthing center, general acute-care hospital, or medical laboratory by reason of having provided the information or material to the Kentucky Birth Surveillance Registry.
- (6) The Department for Public Health may implement the provisions of KRS 211.651 to 211.670 through the promulgation of administrative regulations in accordance with the provisions of KRS Chapter 13A.

**Effective:** July 15, 2002

**History:** Amended 2002 Ky. Acts ch. 287, sec. 1, effective July 15, 2002. -- Amended 1998 Ky. Acts ch. 327, sec. 1, effective July 15, 1998; and ch. 426, sec. 310, effective July 15, 1998. -- Amended 1994 Ky. Acts ch. 405, sec. 77, effective July 15, 1994. -- Created 1992 Ky. Acts ch. 62, sec. 3, effective July 14, 1992.

**211.665 Advisory committee -- Duties.**

- (1) The secretary shall appoint a committee to advise on the implementation of the Kentucky birth surveillance registry. The advisory committee shall have representation from the disciplines of obstetrics and gynecology, pediatrics, genetics, epidemiology, biostatistics, hospital administration, state agency service providers, parents of children with congenital anomalies, or high risk conditions, and consumers. Other disciplines may be represented at the discretion of the secretary.
- (2) The advisory committee shall recommend to the department a definition of congenital anomalies and high risk conditions to be reported to the registry.
- (3) If the department determines it is necessary to collect information from sources other than vital statistics records, general acute care hospitals, medical laboratories, and free-standing birthing centers, the department shall consult with the advisory committee prior to requesting the information.

**Effective:** July 15, 2002

**History:** Amended 2002 Ky. Acts ch. 287, sec. 2, effective July 15, 2002. -- Amended 1994 Ky. Acts ch. 405, sec. 78, effective July 15, 1994. -- Created 1992 Ky. Acts ch. 62, sec. 4, effective July 14, 1992.

**211.670 Confidentiality of registry reports and records -- Use of information.**

- (1) All lists and medical records maintained by hospitals and medical laboratories pursuant to KRS 211.660 shall be confidential. All information collected and analyzed pursuant to KRS 211.660 and 211.665 shall be held confidential as to the identity of the individual patient. Staff of the cabinet, the department, or its designee may use the information to notify parents of available medical care and other services available for the child and family. Further disclosure shall be made only pursuant to the written consent of the child's parent or legal guardian.
- (2) Access to information assembled by the Kentucky birth surveillance registry shall be limited to the cabinet, the department, or its designee and to qualified persons or organizations engaged in demographic, epidemiological or other similar studies related to health and health care provision. A written agreement to maintain confidentiality shall be required if access is approved for persons other than representatives of the cabinet.
- (3) The department shall maintain a record of all persons given access to the information in the Kentucky birth surveillance registry. The record shall include: the name of the person authorizing access; name, title, and organizational affiliation of person given access; dates of access; and the specific purpose for which information is to be used. This record of access shall be open to public inspection during normal operating hours of the department.
- (4) Information assembled by the Kentucky birth surveillance registry may be disclosed in summary, statistical, or other form which does not identify particular individuals or individual sources of information.
- (5) Any person who, in violation of a written agreement to maintain confidentiality, discloses any information provided under KRS 211.660 and 211.665 may be denied further access to confidential information maintained by the department.

**Effective:** July 15, 2002

**History:** Amended 2002 Ky. Acts ch. 287, sec. 3, effective July 15, 2002. -- Amended 1994 Ky. Acts ch. 405, sec. 79, effective July 15, 1994. -- Created 1992 Ky. Acts ch. 62, sec. 5, effective July 14, 1992.

## Appendix 2: Codes Collected- Kentucky Administrative Regulation 902 KAR 19:010

RELATES TO: KRS 211.655, 211.660, 211.670

STATUTORY AUTHORITY: KRS 211.660(6)

NECESSITY, FUNCTION, AND CONFORMITY: KRS 211.660(1) requires the department to establish the Kentucky Birth Surveillance Registry based on the need to provide information on the incidence, prevalence, and trends of congenital anomalies, stillbirths and high risk conditions; provide information as to possible causes; and develop preventive strategies to reduce their incidence and the secondary complications associated with them. This administrative regulation establishes uniform procedures for collection of data for the registry.

Section 1. Definitions. (1) "Agent" means an entity with which the department may:

- (a) Contract pursuant to carrying out the duties of the registry; and
- (b) Designate to act on the behalf of the registry to edit or analyze data from hospitals.

(2) "Cabinet" means the Cabinet for Health Services.

(3) "Coding and transmission specifications" or "UB-92 Submission Manual" means the technical directives the cabinet issues concerning technical matters subject to frequent change, including codes and data for uniform provider entry into particular character positions and fields of the UB-92 and uniform provider formatting of fields and character positions for purposes of electronic data transmissions pursuant to 902 KAR 17:040 or, where not specified, as are delineated in the UB-92 Training Manual.

(4) "Department" means the Department for Public Health.

(5) "Division" means the Division of Adult and Child Health, which is the administrator of the Kentucky Birth Surveillance Registry, and is located within the Department for Public Health.

(6) "Hospital" means an acute care hospital licensed under the provisions of KRS Chapter 216B.

(7) "Hospitalization" means the inpatient medical episode identified by a patient's birth, admission date, length of stay and discharge date, and further identified by a provider-assigned patient control number unique to that inpatient episode.

(8) "Laboratory" means a medical laboratory licensed under KRS Chapter 333.

(9) "ICD-9 Code" means the diagnosis code specifications under the International Classification of Diseases, in current usage, required for reporting diagnoses and diseases to all U.S. Public Health Service and Health Care Financing Administration programs.

(10) "Medical record" means the patient's actual medical record maintained by the hospital's medical record department or by a laboratory.

(11) "Record" means documentation in UB-92 format, in paper or electronic form, of:

- (a) A hospitalization;
- (b) An outpatient visit; or
- (c) A laboratory result.

(12) "Registry" means the Kentucky Birth Surveillance Registry.

(13) "UB-92" means the billing form identified by the Federal Health Care Financing Administration as HCFA Form 1450, as recommended by the National Uniform Billing Committee and adopted by the Kentucky Uniform Billing Committee for use by hospitals and other providers in billing for hospitalizations.

Section 2. Data Collection. (1) Hospitalization records. A hospital shall document, on a UB-92 record, each hospitalization it provides for an inpatient of age five (5) years or under who is diagnosed with a congenital birth anomaly or high-risk condition, as defined by the department in accordance with KRS 211.660(2), and included in Section 7 of this administrative regulation. Each hospital shall provide to the registry the data specified in Section 8 of this administrative regulation.

(2) Outpatient and laboratory records.

(a) In accordance with KRS 211.660(3)(b), a laboratory shall maintain medical records for each person tested who is five (5) years of age or younger and who has a primary diagnosis or laboratory test result indicating a congenital anomaly or high-risk condition, as defined by the department and included in Section 7 of this administrative regulation.

(b) A laboratory, and a hospital voluntarily maintaining an outpatient list as described at KRS 211.660(3)(a), shall provide the data specified in Section 8 of this administrative regulation.

(3) Access to records. A reporting entity shall provide a requesting agent of the registry with access to the medical record of any patient meeting the criteria in subsections (1) or (2) of this section, as authorized by KRS 211.660(4).

Section 3. Data Finalization and Submission. (1) Submission of final data. Data shall be deemed final for purposes of submission to the registry as soon as a record is sufficiently final that the provider could submit it to a payor for billing purposes, whether or not the record has actually been submitted to a payor.

(a) Finalized data shall not be withheld from submission to the registry on grounds that it remains subject to adjudication by a payor; and

(b) Data on a hospitalization shall not be submitted to the registry before:

- 1. The patient is discharged; or
- 2. The record is sufficiently final that it could be submitted to a payor for billing.

(2) Transmission of records.

(a) Data submitted to the registry shall be uniformly completed and formatted according to coding and transmission specifications;

(b) Hospitals and laboratories that have the capacity shall submit data on computer-readable electronic media;

(c) Hospitals and laboratories shall provide backup security against accidental erasure or loss of the data until any incomplete or inaccurate records identified by the registry have been corrected and resubmitted;

(d) Data submitted by mail shall be by certified mail or other traceable carrier, such as United Parcel Service; and

(e) A hospital or laboratory that submits records in the form of paper copies shall either deliver the copies to the registry's reporting agent, or send them in secure packaging by mail postmarked on or before the due date.

Section 4. Data Submission Timetable. Quarterly submission. A hospital shall submit data at least once for each calendar quarter. A quarterly submission shall contain data from records of patients which became final during that quarter, as specified in Section 3(1) of this administrative regulation. The data shall be submitted to the registry not later than forty-five (45) days after the last day of that quarter.

(1) If the 45th day falls on a weekend or holiday, the submission due date shall become the next following working day.

(2) Outpatient data and laboratory reports shall be submitted directly to KBSR within thirty (30) days of the written request.

(3) A hospital shall, within thirty (30) days of receipt of a written request from the registry, submit a medical records report for specified ICD-9 codes for a designated quarter.

Section 5. Data Corrections. (1) Editing. The following UB-92 data fields from Section 9 of this administrative regulation shall be edited by the registry upon receipt, in order to ensure completeness and validity of the data for further processing: patient name, insured's name.

(2) If the registry identifies a record as incomplete or invalid, the submitting hospital shall submit a corrected copy within thirty (30) days of notification. Date of notification shall be considered to be the date postmarked on the registry's mailed notice of required correction. Submission shall be by either electronic transmission or mailing.

Section 6. Working Contacts. (1) Beginning January 1, 1996 and annually thereafter, each hospital required to submit data shall report, by letter to the registry, the names and telephone numbers of a chief executive officer shall not be designated as a contact or backup, unless no other employee has the required technical expertise.

(2) If the chief executive officer, designated contact person or back-up person changes during the year, the name of the replacing person shall be reported immediately to the registry.

Section 7. Required Reporting Conditions. The data which are submitted from the hospital to the registry shall be at least for those patients, from birth to five (5) years of age, for whom any reported diagnoses includes the following ICD-9 codes:

(1) All congenital anomalies codes - 740-759. (Examples: microcephaly 742.1; macrocephaly 742.4; upper GI anomalies 750; lower GI anomalies 751; gastroschisis/omphalocele 756.7; chromosome anomalies 758.)

(2) Dwarfism not elsewhere classified - 259.4.

(3) Metabolic/storage disorders - 270-279, Excluding codes 274, 276 and 278.

(4) Hereditary hemolytic anemia - 282.

(5) Neurologic disorders of brain and spinal cord - 334-335.

(6) Cerebral palsy - 343.

(7) Teratogens (noxious influences) - 760.7 and all subcategories, from 760.70 to 760.79.

(8) Infant of diabetic mother - 775.0.

(9) Failure to thrive - 783.4.

(10) Small for gestational age - 764.0.

Section 8. Required Data Elements. (1) UB-92 data. Hospitals shall ensure that each copy of UB-92 data submitted to the registry contains at least the following data elements as provided on the UB-92 form. Asterisks identify elements that shall not be blank and shall conform to coding and transmission specifications.

UB-92 FIELD #	ELEMENT NAME
*5	Federal Tax Number
*12	Patient Name
13	Patient Address
*14	Birth Date
*15	Sex
*17	Admission/Start of Care Date
*23	Medical Record #
*58	Insured's Name
59	Patient Relationship
60	Certificate/SSN/Health Insurance Claim/ID Number
*67	Principal Diagnosis Code
68-75	Other Diagnosis Codes (Up to 8)
*82	Attending Physician Unique Identification Number or Alternate Number

(2) Outpatient and laboratory data. A laboratory and a hospital voluntarily maintaining a list of outpatients, in accordance with KRS 211.660(3)(a), shall ensure that the data submitted to the registry includes the following data elements: patient name, patient address, birth date, sex, principal diagnosis, other diagnoses (up to eight (8)), and reporting source.

Section 9. Incorporation by Reference. (1) "UB-92 Training Manual (August 4, 1993)" is incorporated by reference.

(2) This material may be inspected, copied, or obtained, subject to applicable copyright law, at the Kentucky Birth Surveillance Registry, Division of Adult and Child Health, Department for Public Health, 275 East Main Street, Frankfort, Kentucky 40621, Monday through Friday, 8 a.m. to 4:30 p.m. (22 Ky.R. 1185; Am. 1480; 1604; eff. 3-7-96; 29 Ky.R. 574; 966; eff. 10-16-2002.)

### Appendix 3: Codes Abstracted- Revised March 2016

Birth Defects	ICD-9-CM Codes	ICD-10-CM Codes
<b>Central Nervous System</b>		
Anencephaly	740.0 – 740.1	Q00.0, .1
Spina bifida without anencephaly	741.0, 741.9	Q05.0-Q050.9, Q07.01, .01
Microcephaly	742.1	Q02
Encephalocele	742.0	Q01.0-Q01.9
Holoprosencephaly	742.2	Q04.1-Q04.4
<b>Eye</b>		
Anophthalmia/microphthalmia	743.0, 743.1	Q11.0-Q11.2
Congenital cataract	743.30 – 743.39	Q12.0, .1, .3, .4, .8, .9
<b>Ear</b>		
Anotia/microtia	744.00-744.09	Q16.0, .1, .3, .4, .5, .9
Anotia/microtia	744.23	Q17.2
<b>Cardiovascular</b>		
Common truncus (truncus arteriosus or TA)	745.0	Q20.0
Transposition of the great arteries (TGA)	745.10, .11, .12, .19	Q20.1, .3, .5, .8
Tetralogy of Fallot (TOF)	745.2	Q21.3
Single ventricle	745.3	Q20.4
Interrupted /Hypoplastic aortic arch	747.11, 747.22	Q25.2
Endocardial cushion defect	745.60, .61, .69	Q21.2
Pulmonary valve atresia and stenosis	746.01, 746.02	Q22.0, .1
Tricuspid valve atresia and stenosis	746.1	Q22.4
Ebstein anomaly	746.2	Q22.5
Aortic valve stenosis	746.3	Q23.0
Hypoplastic left heart syndrome	746.7	Q23.4
Coarctation of aorta	747.1	Q25.1
Total anomalous pulmonary venous connection	747.41	Q26.2
<b>Orofacial</b>		
Cleft palate alone (without cleft lip)	749	Q35.1, .3, .5, .7, .9
Cleft lip alone (without cleft palate)	749.1	Q36.0, Q36.9
Cleft lip with cleft palate	749.20-749.25	Q37.0-.5, .8, .9
Choanal atresia	748.0	Q30.0

**Gastrointestinal**

Esophageal atresia/tracheoesophageal fistula	750.3	Q39.0-.4
Rectal and large intestinal atresia/stenosis	751.2	Q42.9
Biliary atresia	751.61	Q44.2, .3
Small intestinal atresia/stenosis	751.1	Q41.9
Pyloric stenosis	750.5	Q40.0
Hirschsprung disease (congenital megacolon)	751.3	Q43.3

**Genitourinary**

Renal agenesis/hypoplasia	753.0	Q60.2, Q60.5
Bladder exstrophy	753.5	Q64.10, .19
Congenital posterior urethral valves	753.6	Q64.2, .31, .32, .33, .39

**Musculoskeletal**

Limb deficiencies (upper limb)	755.2	Q71.1, .4, .5, .899, .9
Limb deficiencies (lower limb)	755.3	Q72.1, .3, .4, .5, .6, .7, .899
Limb deficiencies (unspecified limb)	755.4	Q73.0, .1, .8
Gastroschisis	756.73	Q79.3
Omphalocele	756.72	Q79.2
Diaphragmatic hernia	756.6	Q79.0, .1

**Chromosomal**

Trisomy 13	758.1	Q91.7
Trisomy 21	758.0	Q90.9
Trisomy 18	758.2	Q91.3
Autosomal deletion syndromes	758.31 - 758.39, 758.5	Q93.0-Q93.9
Turner syndrome	758.6	Q96.0-Q96.9
Other chromosomal syndromes	758.81, .89, .9	Q99.8, Q99.9

**Other**

Syndromes affecting multiple systems	755.55, 759.81	Q87.0, .1, .2, .3, .5, .81, .89
Other unspecified congenital anomalies	759.89	E78.71, .72
Fetus/ newborn affected by maternal substance use	760.70, .71, .72, .73, .75, .79	P04.2, .3, .41, .49, .8, .9; Q86.0, Q86.8