Running the Numbers

A Periodic Feature to Inform North Carolina Health Care Professionals About Current Topics in Health Statistics

Critical Congenital Heart Defects in North Carolina

Congenital heart defects are the most common type of serious birth defect, occurring in nearly 40,000 infants (approximately 1% of all live births) each year in the United States [1]. Such defects are a leading cause of infant death, accounting for at least 25% of infant deaths related to birth defects in the United States [2-4]. In North Carolina during the 5-year period 2005-2009, 7,690 infants were diagnosed with congenital heart defects, or about 1 in every 83 infants; approximately 10% of these infants died within the first year of life [North Carolina Birth Defects Monitoring Program, unpublished data, 2012].

In recent years there has been a decline in infant and childhood mortality from congenital heart defects in the United States, due in large part to improvements in diagnosis, surgical management, and catheter-based interventions [5]. In many cases, the key to improved survival and reduced risk of serious disability depends on early detection, often before the newborn is discharged from the hospital. Recognizing the importance of early detection, the US secretary of Health and Human Services, Kathleen Sebelius, in September, 2011, adopted recommendations of the secretary's Advisory Committee on Heritable Disorders in Newborns and Children that a subgroup of 7 specific types of defects known as critical congenital heart defects (CCHD) be added to the recommended universal screening panel for newborns, using pulse oximetry screening. The conditions targeted for pulse oximetry screening are hypoplastic left heart syndrome, pulmonary atresia with intact ventricular septum, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus (common truncus) [6]. In each case, the two atria and two ventricles (or in lay terms, the receiving and pumping chambers) of the normal heart are atretic, mislocated, unconnected, or blocked so blood does not flow normally from the right side of the heart through the lungs to the left, and blood is poorly oxygenated. These 7 conditions may also be classified as cyanotic congenital heart defects, because the affected infant often presents with hypoxemia within the first few days or weeks of life.

Infants with CCHD who are not diagnosed promptly are at high risk for cardiovascular shock and subsequent death when the ductus arteriosus closes and prevents blood from flowing through this connection that had allowed oxengated blood to mix into the circulation. Although CCHD may be diagnosed through prenatal ultrasound or by physical examination, those methods are unreliable and can result in missed diagnoses. Screening with pulse oximetry, which is a simple noninvasive test of blood-oxygen saturation, can significantly improve detection of CCHD and reduce the risk of death and serious disability. Pulse oximetry screening can identify asymptomatic infants with CCHD within 24-48 hours of birth, providing an opportunity for prompt referral to a specialist to provide critical medical care needed to prevent death or serious disability. The screening can also detect other life-threatening medical conditions that can lead to low blood-oxygen levels, including sepsis, pneumonia, or pneumothorax. The secretary's Advisory Committee on Heritable Disorders in Newborns and Children, in collaboration with the American Academy of Pediatrics, the American College of Cardiology, and the American Heart Association, has developed a screening algorithm to help facilitate implementation of universal screening for CCHD by states [7]. The screening

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TABLE 1.
Prevalence of Critical Congenital Heart Defects (CCHD) in North Carolina, 2005-2009

Phenotype	Number of Cases[a]	Prevalence (per 10,000 live births)	95% CI[b]
Truncus arteriosus	56	0.88	(0.66, 1.14)
Transposition of great arteries	160	2.50	(2.13, 2.92)
Tetralogy of Fallot	256	4.01	(3.53, 4.53)
Pulmonary valve atresia with intact septum	100	1.56	(1.27, 1.90)
Tricuspid valve atresia	109	1.71	(1.40, 2.06)
Hypoplastic left heart syndrome	169	2.64	(2.26, 3.07)
Total anomalous pulmonary venous return	81	1.27	(1.00, 1.57)
Total Number of Infants with CCHD	828	12.96	(12.09, 13.87)

[a]Some infants had more than one type of CCHD; therefore the numbers for each phenotype when added together do not equal the total number of infants with CCHD.
[b]CI, confidence interval

algorithm was developed to improve case detection with a very low rate of false positives (ie, specificity >99.5%). They recommend a broad-based implementation strategy, involving birth facilities, pediatric cardiac surgery centers, primary care providers, and state public health programs such as Title V maternal and child health programs and state-based birth defect surveillance systems. The role of public health in this model is to provide monitoring, quality assurance, and evaluation of universal screening for CCHD.

The purpose of this Running the Numbers article is to provide baseline data on the prevalence and characteristics of infants with CCHD in North Carolina. This information can be used to assist with the initial planning and implementation of universal screening for CCHD, and to help assess the public health impact of such screening, including mortality and economic outcomes.

Methods

Data for this report are from the North Carolina Birth Defects Monitoring Program and cover infants who were born during the 5-year period of 2005-2009. The Birth Defects Monitoring Program is a statewide, population-based, active surveillance system that has been in operation since 1995. The program is housed within the State Center for Health Statistics in the Division of Public Health, North Carolina Department of Health and Human Services. Under state law, the program has the authority to obtain from hospitals and licensed physicians in the state all information on infants suspected of having a congenital malformation, and to review and abstract that information. All patient-identifying information collected and maintained by the program is considered confidential by state law. The case definition includes all infants born to a mother residing in North Carolina who were diagnosed with a major structural malformation in the first year of life, as well as all fetuses in which malformations were diagnosed after fetal death or pregnancy termination. Birth Defects Monitoring Program field staff review approximately 12,000 medical charts annually using a multistage process involving initial case finding followed by medical record review and abstraction to confirm suspected diagnoses. Information in the medical record that is reviewed includes discharge summaries; prenatal test results; labor and delivery information; newborn and pediatric examinations; diagnostic findings from magnetic resonance imaging, echocardiography, or other imaging studies; cytogenetic and laboratory findings; surgical reports; pathology records; and autopsy results. All data from live-born infants and fetal deaths are linked back to the infant's or fetus' vital record in order to obtain additional demographic data and to facilitate calculation of rates.

Results

During the 5-year period 2005-2009, there were 828 cases of CCHD, of which 795 (96%) were live-born infants and 33 (4%) were fetal deaths or pregnancy terminations. The overall prevalence of CCHD was 12.96 per 10,000 live births; 56% of those affected had 1 or more additional, extracardiac malformations, and 44% of the infants

TABLE 2.
Characteristics of Infants with Critical Congenital Heart Defects (CCHD) and Infants Without Birth Defects, and of Their Mothers, in North Carolina, 2005-2009

Characteristic	Infants with CCHD (N = 828) No.[a] (%)	Infants without Major Birth Defects (N = 620,140) No.[a] (%)
Gender of infant		
Male	463 (56.0)	315,822 (50.9)
Female	364 (44.0)	304,314 (49.1)
Birth weight		
<1,500 grams	53 (6.5)	9,579 (1.5)
1,500-2,499 grams	166 (20.4)	43,831 (7.1)
≥2,500 grams	596 (73.1)	566,516 (91.4)
Gestational age (weeks)		
<32	50 (6.1)	10,957 (1.8)
33-36	148 (18.0)	54,114 (8.7)
≥37	626 (76.0)	554,793 (89.5)
Maternal age (years)		
< 20	100 (12.1)	71,696 (11.6)
20-24	209 (25.2)	165,316 (26.7)
25-29	230 (27.8)	169,315 (27.3)
30-34	171 (20.6)	135,785 (21.9)
≥35	118 (14.3)	77,996 (12.6)
Maternal Race/ethnicity		
White/non-Hispanic	456 (55.1)	345,295 (55.7)
Black/non-Hispanic	200 (24.2)	145,203 (23.4)
Hispanic	127 (15.3)	101,805 (16.4)
Asian/Pacific Islander	27 (3.3)	18,615 (3.0)
Native American	15 (1.8)	8,352 (1.4)
Other/unknown	3 (0.4)	870 (0.1)
Mother's no. of previous live births		
None	321 (39.2)	251,760 (40.6)
1-2	382 (46.7)	305,241 (49.3)
≥3	115 (14.1)	62,602 (10.1)
Delivery paid by Medicaid		
Yes	434 (54.6)	301,069 (48.6)
No	361 (45.4)	319,071 (51.4)
Level of care at delivery hospital		
Level III (tertiary)	508 (61.4)	252,413 (40.7)
Other (nontertiary)	320 (38.6)	367,727 (59.3)
Infant death		
Yes	215 (26.0)	3,845 (0.6)
No	613 (74.0)	616,295 (99.4)

[a]Numbers shown for each characteristic may not add up to total number of infants in category because information is missing for some individuals.

had isolated CCHD. Table 1 shows the prevalence of each CCHD phenotype (ie, type of CCHD). Tetralogy of Fallot was the most commonly occurring CCHD, with a prevalence of 4.01 per 10,000 live births, affecting 256 (30.9%) of all children diagnosed with CCHD. Hypoplastic left heart syndrome was the second-most-common type, with a prevalence of 2.50 per 10,000 live births. Truncus arteriosus was the least common defect, affecting only 56 infants during the 5-year period.

TABLE 3.
Medicaid Claims During the First Year of Life for Infants Diagnosed with Critical Congenital Heart Defects (CCHD) in North Carolina, 2005-2009

Group	No. of Infants	Total Value of Claims	Median Value of Each Child's Claim
All infants with CCHD who were covered by Medicaid	462	$53.4 million	$72,466
Infants with isolated CCHD	189	$15.0 million	$57,610
Infants with nonisolated CCHD	273	$38.4 million	$86,560
Infants who died	128	$17.3 million	$92,316
Infants who survived	334	$36.2 million	$71,151

Table 2 presents maternal and infant characteristics in cases of CCHD, compared with the characteristics of infants without major birth defects and their mothers. The infants with CCHD were more likely to be male, to be born preterm (<37 weeks), and to weigh less than 2,500 grams. Compared with mothers of infants without birth defects, mothers of infants with critical congenital heart defects were more likely to have had 3 or more previous live births, to have had their delivery paid for by Medicaid, and to have delivered in a tertiary (level III) hospital. The risk of death within the first year of life among infants with CCHD was substantially higher than that among infants without birth defects (26.0% versus 0.6%). Deaths among infants with CCHD were evenly distributed between the neonatal (49%) and postneonatal (51%) period. Low birth weight and low gestational age were important risk factors for infant death among the infants with CCHD, as was high parity of the mother.

Approximately 56% (462) of the 828 infants with CCHD had Medicaid coverage during their first year of life (Table 3). Medicaid claims for these 462 infants during that year totaled $53.4 million, and the median claim value per child was $72,466. The median claim value for an infant with nonisolated CCHD was 1.5 times that for an infant with CCHD only. The median claim value for an infant who died was $92,316—about 30% higher than that for an infant who survived.

Conclusion

Despite medical advances in the treatment of infants with congenital heart disease, mortality among infants with CCHD in North Carolina remains high, particularly among those infants who are born prematurely. The prevalence of most CCHD phenotypes in North Carolina is similar to that of pooled national prevalence estimates and published data from individual state-based birth-defect surveillance programs [8, 9]. Given the relatively high prevalence of CCHD in the state (about 1 in 772 newborns), the availability of life-saving interventions, and the importance of early detection to reduce the risk of mortality, North Carolina should take the necessary steps recommended by the US Secretary of Health and Human Services to implement a statewide universal screening program for CCHD using pulse oximetry screening. Even without a legislative requirement for screening, many birth centers in the state have begun screening because it has little upfront costs, is straightforward to implement, and can lead to substantial improvement in health outcomes. NCMJ

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