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# **Birth Defects in North Carolina**

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**A Report by the  
North Carolina Birth Defects Monitoring Program  
State Center for Health Statistics  
Division of Public Health  
Department of Health and Human Services**

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**January 2006**

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A Report by the  
North Carolina Birth Defects Monitoring Program

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**January 2006**

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## Introduction

A birth defect is a structural, functional, or chemical abnormality that is present at birth. Birth defects are a leading cause of infant death and childhood disability. Approximately three percent of all babies, or about one in every 33, are born with serious birth defects. Each year in North Carolina more than 3,500 infants are born with major birth defects. Among the 966 infants who were born in 2003 and died within the first year of life, 263 (27.2 percent) had a diagnosed birth defect.

The economic burden of birth defects in North Carolina is substantial. In 2003, in-patient hospital care for children under age 18 with a primary diagnosis of a birth defect was \$73.4 million, and the average charge per admission was \$16,572. Fifty percent of all infants with birth defects are enrolled in Medicaid during the first year of life. The total first-year Medicaid paid claims for infants with birth defects in 2003 was \$65.1 million. That year, the average Medicaid costs for infants with birth defects was \$27,517, compared to \$20,524 for infants born preterm and \$2,922 for infants born without such conditions.

The causes of most birth defects are unknown, though some have been linked to genetic factors, maternal illnesses, certain medications, and environmental influences. Some birth defects, such as fetal alcohol syndrome and congenital rubella syndrome, are entirely preventable. Research studies have also shown that daily intake of 400 micrograms of the B-vitamin folic acid can greatly reduce the risk for birth defects such as spina bifida and anencephaly.

## North Carolina Birth Defects Monitoring Program

The North Carolina Birth Defects Monitoring Program (NCBDMP) operates under the statutory authority (G.S. 130A-131) of the State Center for Health Statistics (SCHS), North Carolina Department of Health and Human Services. Funding for the NCBDMP is provided by state appropriations and through a cooperative agreement with the

National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention (CDC).

### ***Case Definition, Data Collection, and Confidentiality***

The NCBDMP is a statewide, population-based surveillance system that collects information on all infants in North Carolina who are born with major birth defects. Data are collected by specially trained field staff who review and abstract data from all hospitals that provide labor and delivery and pediatric services, as well as from selected specialty clinics and other facilities throughout the state. Surveillance data are obtained from more than 90 hospitals and medical facilities statewide. The data are maintained in the program's central registry, which is a confidential electronic database housed within the State Center for Health Statistics. In order to be included in the registry, the infant must have been born to a resident of North Carolina and be diagnosed with one or more birth defects within the first year of life. The registry includes all live-born infants, fetal deaths of 20 or more weeks gestation, and pregnancy terminations regardless of gestational age. For live births and reported fetal deaths, residency at the time of birth is verified by matching case records to state vital statistics files, whereas for pregnancy terminations prior to 20 weeks, North Carolina residence is determined from the medical record. The NCBDMP uses the British Pediatric Association (BPA) coding system, which is widely used by state and international birth defect surveillance programs.

All personal identifying information collected and maintained by the NCBDMP is considered confidential by state law. Identifying information may be released for epidemiologic research or public health purposes only, contingent upon approval of the research protocol by the SCHS and by an authorized Institutional Review Board. Published data are presented at an aggregate level in order to protect patient confidentiality. The NCBDMP makes certain aggregate data available to the public through the SCHS web query system at [www.schs.state.nc.us/SCHS/data/query.html](http://www.schs.state.nc.us/SCHS/data/query.html). Please visit this site if you are interested in birth defects data for specific counties or perinatal regions.

## **Program Goals and Objectives**

The purpose of the NCBDMP is to collect, analyze, and disseminate information related to the occurrence, prevention, and treatment of birth defects in North Carolina. This information is used to improve the health status of infants and children in North Carolina in many ways, including:

- Monitoring geographic and temporal trends of birth defects;
- Identifying populations at increased risk, and helping target those populations with public health interventions;
- Evaluating the effectiveness of interventions and services;
- Providing birth defects information to health care providers, researchers, and the public;
- Improving access to services through identification of children with special needs; and
- Engaging in research aimed at understanding the causes of birth defects and identifying potential new avenues for prevention.

## **North Carolina Birth Defects Monitoring Program Collaborative Projects**

The NCBDMP is actively involved in several collaborative projects with other programs and agencies. These collaborations are important in helping the program meet its objectives and fulfill its mandate to serve the public health needs of the state. Two of these projects are described below.

### ***Neural Tube Defect Recurrence Prevention Program***

The Neural Tube Defect (NTD) Recurrence Prevention Program is a collaborative activity involving the NCBDMP, the Genetics and Newborn Screening Unit, and the Early Intervention Program in the Division of Public Health. The primary goal is to reduce the risk of subsequent NTDs among women with a previously affected pregnancy, by educating

them and their healthcare providers about the need for a higher dose of folic acid for such women. Women with a previous NTD pregnancy should consume 4 milligrams of folic acid per day, beginning prior to planning a subsequent pregnancy and continuing through the first trimester. This amount of folic acid is available only by a doctor's prescription. Another purpose of the program is to provide information to the family concerning medical and financial services that are available to assist them in caring for their child. Initiated in 1999 through a cooperative agreement with CDC, the program is now maintained as an ongoing activity within the division.

### ***North Carolina Center for Birth Defects Research and Prevention***

The North Carolina Center for Birth Defects Research and Prevention is a collaborative effort between the North Carolina Birth Defects Monitoring Program and the Department of Epidemiology at the University of North Carolina at Chapel Hill. North Carolina is one of nine such centers across the country. Each center's goal is to conduct epidemiologic research for the prevention of birth defects. A major focus of each center is to participate in the National Birth Defects Prevention Study (NBDPS), which is the largest ongoing case-control study of birth defects ever conducted. Women participating in this study provide information about risk factors such as diet, medications, and pregnancy history through a detailed telephone interview. Participants are also asked to provide cheek cells from the infant and parents so that DNA can be studied to identify possible genes that may be associated with certain birth defects. All of this information is important in helping scientists understand the causes of birth defects, which is an essential step in identifying new approaches for prevention. As a part of this project, the North Carolina Center is conducting an evaluation of Medicaid expenditures and health care service utilization among children with cleft lip and cleft palate. This evaluation is also examining potential barriers to care which, if eliminated, may improve access to needed services for these families and children.

## Technical Notes

Unless otherwise indicated, all data in this report are based on infants born during calendar year 2003. There were 118,292 North Carolina resident live births in that year. Prevalence data shown in the appendix of this report are calculated using the following formula:

$$\text{Prevalence} = (\text{number of cases} / \text{total number of live births}) \times 10,000$$

Pregnancy terminations or fetal deaths occurring prior to 20 weeks gestation are not included in the data shown in the appendix. As a general rule, prevalence data based on fewer than 10 observed cases tend to be unreliable; that is, they are less likely to reflect the true prevalence than are those based on a larger number of cases. The reader is advised to use caution when interpreting statistical information based on small numbers of events. The degree of precision or certainty of a prevalence estimate is reflected by the width of the confidence interval, with a wider interval indicating less precision. The table in the appendix provides 95 percent confidence intervals to facilitate interpretation. These confidence intervals are based on the exact binomial limits.

## Overview of Selected Birth Defects

There are several hundred types of birth defects, most of which also occur in many different variants and forms. A comprehensive description of the many types of birth defects is beyond the scope of this report. The following section presents a summary of a few specific birth defects which are relatively common or are of particular interest in North Carolina. Readers interested in learning more about these or other conditions may refer to the Resource list at the end of this report.

## Central Nervous System Defects

The central nervous system (CNS) includes the brain, spinal cord, and associated structures. Birth defects of the central nervous system account for about nine percent of birth defects among infants. Approximately one in every 280 infants is born with a CNS malformation in North Carolina.

Neural tube defects (NTDs), which include anencephaly, spina bifida, and encephalocele, are a type of CNS defect in which the neural tube – the embryonic structure that forms the brain and spinal cord – fails to develop properly during the fourth week of pregnancy. These are very severe conditions, often resulting in death or some degree of physical and neurological impairment. The severity of an NTD depends on its type and location.

**Anencephaly** occurs when the cranial portion of the neural tube fails to close, resulting in incomplete development of the cranium and brain (Figure 1). Infants with anencephaly are unable to survive outside of the womb. Among live births and fetal deaths >20 weeks gestation in North Carolina, about one in 4,000 has anencephaly. Because the malformation is lethal, many additional affected pregnancies are also identified prenatally and terminated before 20 weeks.



Figure 1. Anencephaly

**Spina bifida** occurs when the neural tube fails to close along a portion of the spine, leaving the spinal cord and its membranes exposed (Figure 2). The severity of complications depends upon the size and location of the lesion. If the defect is high on the spinal column, total paralysis of the lower limbs may occur, while a defect at the base of the spine may result in a lesser degree of paralysis. The opening must be surgically repaired soon after birth. Individuals with spina bifida often have associated conditions such as hydrocephaly, clubfoot, mental retardation, muscle weakness/paralysis, loss of bladder and bowel control, as well as other complications. Many are dependent upon wheelchairs, braces, or crutches and typically require lifelong medical care. About one in every 1,970 infants in North Carolina is born with spina bifida.

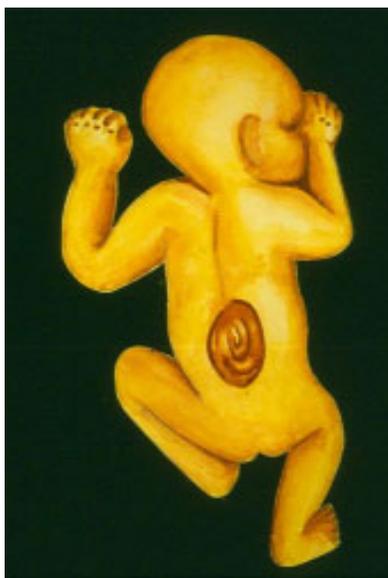


Figure 2. Spina bifida

**Encephalocele** is a herniation of brain tissue through a defect (hole) in the cranium (Figure 3). The location of the defect may be occipital (lower back of the head), posterior parietal (rear side of the skull) or anterior (front part of the skull). Treatment involves surgical closure of the defect and drainage of the cerebrospinal fluid. Mortality rates depend on the location and size of the defect. Infants who survive may have associated disabilities including

hydrocephaly, paralysis, blindness, seizures, mental retardation, retarded growth, and poor muscular coordination. Encephalocele is the least common type of NTD, affecting about one in every 10,750 infants in the state.

In recent years there has been a decline in NTDs in North Carolina as well as nationwide. This decline is believed to be due, in large part, to public health efforts to promote folic acid intake. Studies have shown that daily consumption of folic acid, beginning at least two to three months prior to conception and continuing throughout pregnancy, decreases a woman's risk for having an NTD-affected pregnancy by up to 70 percent. ***Because NTDs occur so soon after conception and because most pregnancies are unplanned, it is essential that all women of childbearing age take a daily multivitamin containing 400 micrograms of folic acid, in addition to eating a balanced, healthy diet.***



Figure 3. Encephalocele

## Cardiovascular Defects

Cardiovascular defects include malformations of the heart and circulatory system. They are the most common type of congenital anomaly, accounting for about 36 percent of all birth defects, and affecting one in every 70 infants born in North Carolina each year.

Between the fifth and eighth weeks of pregnancy, the embryonic structure forming the heart undergoes a process of folding, remodeling, and septation that transforms it into the four chambers of the heart. There are numerous types of cardiovascular defects that may occur during this time. Many factors contribute to these defects. Most cardiac abnormalities seem to be multifactorial (a combination of the environment and the individual's genetic makeup). Some defects, however, can be linked to single-gene mutations, chromosomal abnormalities, and known teratogens – viruses, drugs, and other agents that cause fetal malformations. Cardiovascular defects are often associated with other birth defects and chromosomal anomalies. Treatment of cardiovascular malformations depends on the particular defect and its severity. Minor cardiovascular defects may resolve on their own without medical intervention, while more serious defects often require surgery.

The basic anatomic features of a normal heart are shown in Figure 4. The heart consists of four chambers: the right atrium (RA), left atrium (LA), right ventricle (RV) and left ventricle (LV). After birth, deoxygenated blood from the body enters the right atrium and passes into the right ventricle where it is pumped through the pulmonary artery (PA) and into the lungs to receive oxygen. Oxygenated blood from the lungs then returns through the pulmonary veins into the left atrium, and then passes into the left ventricle where it is pumped out through the aorta (AO) and back into the body.

Because of the heart's complexity, many different types of malformations can occur. Cyanotic heart defects are a group of malformations in which the blood that is pumped from the heart to the body contains an inadequate amount of oxygen. Symptoms include cyanosis, or a bluish color in the infant. Obstructive defects restrict or block the flow of blood from the heart to the body. Septal defects

are abnormal openings or “holes” between the two sides of the heart, allowing blood to flow between the left and right chambers. Infants often have more than one type of cardiac defect at the same time.

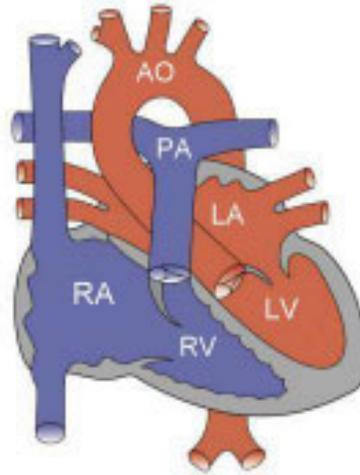


Figure 4. Normal heart

**Atrial septal defects (ASDs)** and **ventricular septal defects (VSDs)** are the most commonly occurring cardiac anomalies, each affecting about one in every 230 infants in North Carolina. Both are often associated with other cardiac defects. An ASD is an abnormal opening in the wall (septum) separating the left and right atria (Figure 5). This defect allows oxygenated blood to flow back into the right side of the heart and to the lungs, instead of being pumped to the body. Many affected infants are asymptomatic, but the increased blood flow to the right atrium

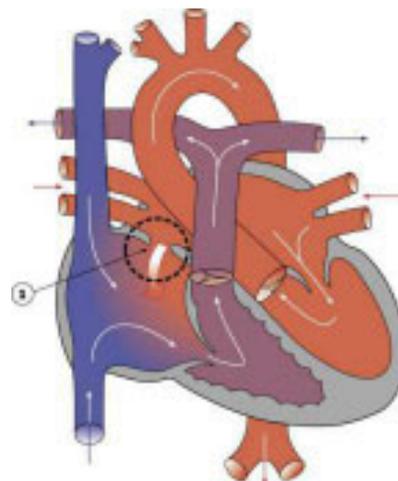


Figure 5. Atrial septal defect (1. hole (defect) in atrial septum)

may result in enlargement of the right ventricle and pulmonary trunk, leading eventually to cardiac failure. Small ASDs may close spontaneously, but more severe cases may require surgical correction.

A VSD is an abnormal opening in the septum separating the left and right ventricles (Figure 6). This hole allows the blood to flow directly from the left to right ventricle, mixing oxygenated with deoxygenated blood which is then carried to the lungs. Symptoms may include congestive heart failure, rapid breathing, and failure to thrive. Like ASDs, the severity of VSDs varies widely depending on the size and number of openings. In mild cases, there may be spontaneous closure while in more severe cases, surgery is required.

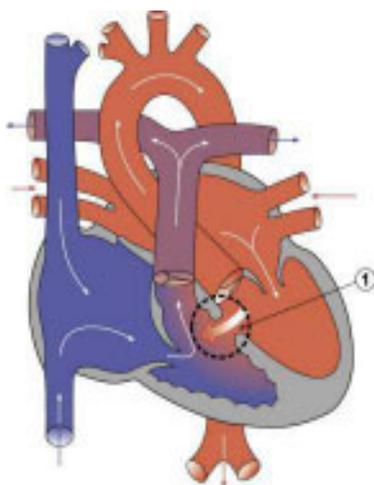


Figure 6. Ventricular septal defect  
(1. hole (defect) in ventricular septum)

**Transposition of the great arteries (TGA)** is a cyanotic heart defect in which the pulmonary artery and the aorta are connected to the wrong ventricles (Figure 7). In TGA, the pulmonary artery connects to the left ventricle instead of the right, and the aorta connects to the right ventricle instead of the left, resulting in the left ventricle emptying into the pulmonary circulation and the right ventricle emptying into the systemic circulation. Symptoms include cyanosis, failure to thrive, and congestive heart failure. Sometimes, infants may have certain other heart defects, such as a patent ductus arteriosus or an atrial septal defect that can reduce the

severity of the symptoms by allowing more oxygenated blood to be pumped to the body. Surgical correction of TGA is required. One in 1,880 infants in North Carolina is affected.

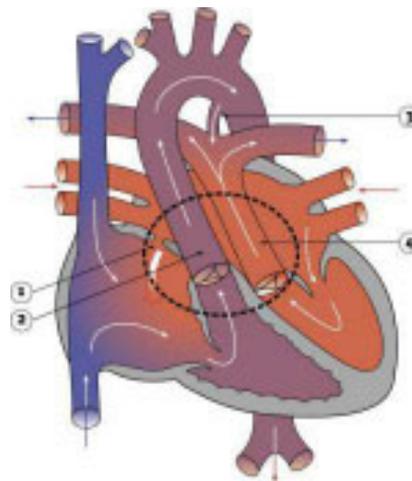


Figure 7. Transposition of Great Arteries  
(1. atrial septal defect; 2. aorta connecting to R. ventricle; 3. patent ductus arteriosus; 4. pulmonary artery connecting to L. ventricle)

**Tetralogy of Fallot**, another type of cyanotic heart defect, is composed of four separate malformations: a ventricular septal defect, hypertrophy of the right ventricle, a malpositioned (overriding) aorta, and stenosis of the pulmonary artery (Figure 8). Infants with this defect show signs of cyanosis, feeding difficulties, and failure to thrive. Prognosis is poor without corrective surgery. In North Carolina, about one in 2,690 infants is born with this malformation.

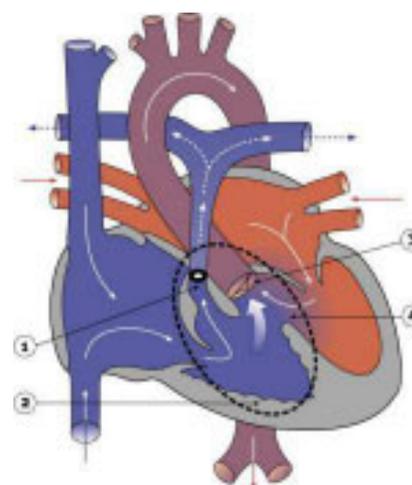


Figure 8. Tetralogy of Fallot  
(1. pulmonary stenosis; 2. R. ventricular hypertrophy; 3. overriding aorta; 4. ventricular septal defect)

**Coarctation of the aorta** is a constriction or pinching of the aorta resulting in decreased systemic blood flow (Figure 9). The coarctation may occur either proximal to (before) or, more commonly distal to (after) the junction of the ductus arteriosus. Affected infants may experience congestive heart failure, but some infants are asymptomatic. Corrective surgery is usually performed. Approximately one in every 1,710 infants in the state is diagnosed with coarctation.

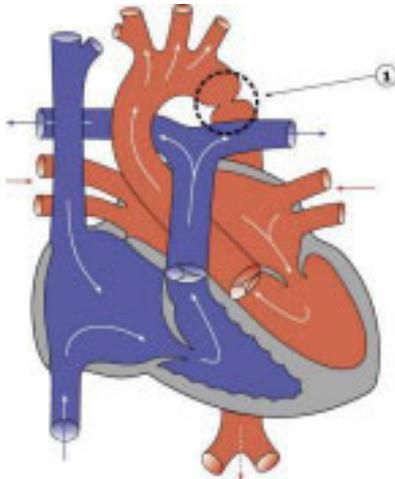


Figure 9. Coarctation of the Aorta  
(1. coarcted or constricted aorta)

**Hypoplastic Left Heart Syndrome (HLHS)** is a complex heart defect in which the left side of the heart is severely underdeveloped (Figure 10). HLHS typically consists of a severely hypoplastic or underdeveloped left ventricle, a hypoplastic aorta, and anomalies of the aortic and mitral valves. Coarctation of the aorta is also frequently found in these infants. In HLHS, oxygenated blood from the lungs flows into the left atrium, and then passes into the right atrium through an opening in the atrial septum (foramen ovale), where it is mixed with deoxygenated blood in the right ventricle. Here, the blood is pumped out through the pulmonary artery, where it eventually reaches the aorta through an opening called the ductus arteriosus. Infants often appear healthy at birth, but quickly become very sick after the ductus closes about the second or third day of life. Symptoms include difficulty with breathing, feeding, and failure to thrive. Surgical repair is sometimes attempted, but prognosis is typically poor. HLHS affects about one in every 4,550 newborns in North Carolina.

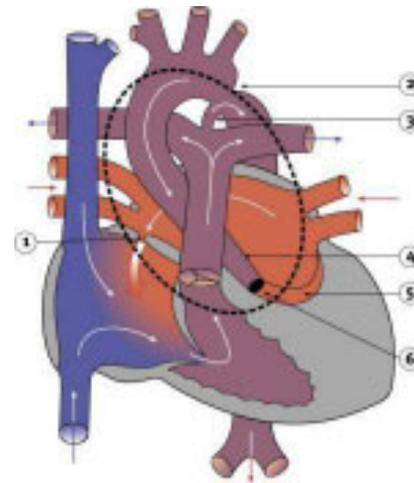


Figure 10. Hypoplastic Left Heart Syndrome  
(1. patent foramen ovale; 2. coarctation of aorta;  
3. patent ductus arteriosus; 4. aortic hypoplasia;  
5. hypoplastic L. ventricle; 6. aortic stenosis)

## Orofacial Clefts

Orofacial clefts, which include cleft palate and cleft lip, occur when the structures of the mouth fail to develop properly. This occurs between the fourth and ninth weeks of pregnancy. Cleft palate and cleft lip may occur individually or together. Approximately 40 percent of orofacial clefts involve clefts of the palate only, while 60 percent involve cleft lip with or without cleft palate. Cleft lip with or without cleft palate is more common in males. Orofacial clefts affect about one in every 670 infants born in North Carolina.

Orofacial clefts can occur alone (isolated or nonsyndromic) or with other birth defects (syndromic). More than one-half of infants with cleft palate (without cleft lip) have one or more additional birth defects; oftentimes these are chromosomal abnormalities. About one-third of infants with cleft lip, with or without cleft palate, have additional birth defects.

**Cleft lip** results from an incomplete closure of the primary palate, which forms the lip and gum (Figure 11). Closure typically takes place by the 45<sup>th</sup> day of pregnancy. **Cleft palate** occurs when there is incomplete closure of the secondary palate, which forms the roof of the mouth (Figure 12). Closure usually takes place around the ninth week of pregnancy.

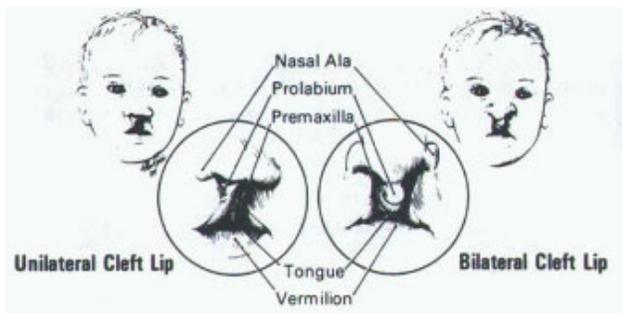


Figure 11. Cleft lip

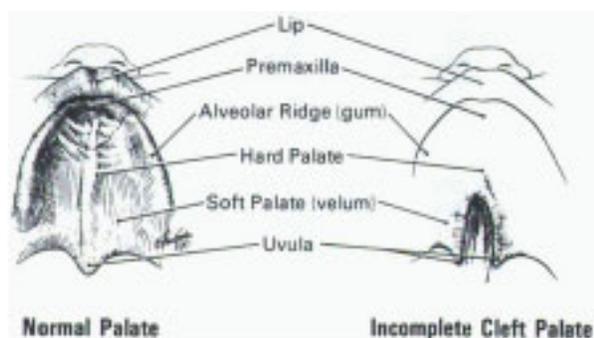


Figure 12. Cleft palate

Cleft palate and cleft lip involve somewhat different processes. When they are both present, it is thought that the clefing of the primary palate interfered with the closure of the secondary palate. Both conditions may create problems with eating, drinking, hearing, and/or speech. Individuals with cleft palate and/or cleft lip are often treated with surgery, orthodontia, and speech therapy.

Some studies suggest that a woman who smokes during pregnancy may have an increased risk for delivering an infant with an orofacial cleft, and that genetic factors play an important role in determining the actual level of risk associated with smoking. In addition, intake of folic acid (400 micrograms/day) before and during pregnancy may prevent some orofacial clefts from occurring.

## Gastrointestinal System Defects

Congenital anomalies involving the gastrointestinal system include those affecting the digestive tract (esophagus, stomach, intestines) and certain organs such as the gallbladder and liver. These anomalies account for about 13 percent of birth defects, and affect one in every 190 infants in the state.

*Tracheoesophageal fistula* is an abnormal opening between the trachea and esophagus allowing liquid and food to enter the lungs (Figure 13). This malformation is frequently associated with *esophageal atresia*, where the esophagus ends in a blind pouch and does not connect with the stomach. Symptoms include respiratory infections and feeding difficulties. Corrective surgery is required. About one in every 3,290 infants in North Carolina is affected with this malformation.

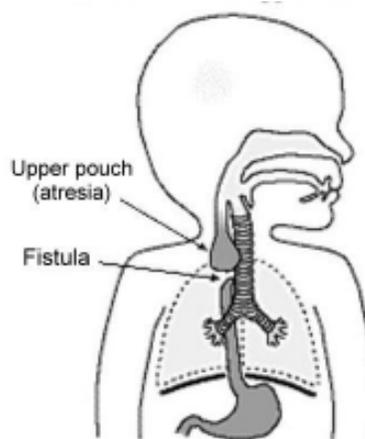


Figure 13. Tracheoesophageal fistula with esophageal atresia

*Pyloric stenosis* is a narrowing of the pylorus, the opening between the stomach and small intestine (Figure 14). This condition prevents food and liquids from passing into the intestines. Infants with pyloric stenosis are asymptomatic at birth, but at about one month of age they begin to experience projectile vomiting and other problems such as weight loss and constipation. Prognosis is excellent after surgical repair. Pyloric stenosis is a relatively common condition, affecting one in every 480 infants in North Carolina.

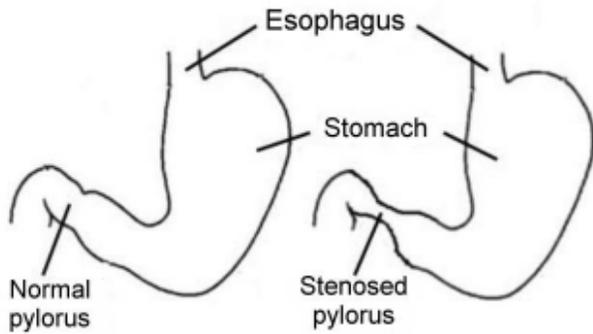


Figure 14. Pyloric stenosis

### Genitourinary Defects

Genitourinary defects include anomalies affecting the internal and external reproductive organs, kidneys, ureters, bladder, and urethra. About one percent of all infants in North Carolina are born with a genitourinary malformation.

**Hypospadias** is a relatively common urinary tract defect in males, in which the urethral opening is located on the underside of the penis (Figure 15). The location of this opening may occur anywhere in the penile/scrotal region, and the severity of the malformation is largely dependent upon the site of the defect. Mild, or first degree, hypospadias is fairly common and is often of no clinical significance, whereas more severe cases (second and third degree) are surgically repaired. About one in every 150 male infants is affected.

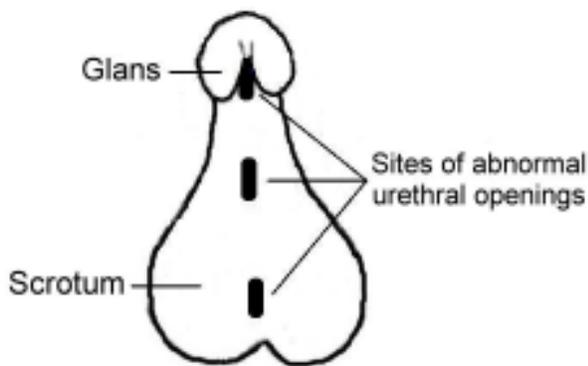


Figure 15. Hypospadias

**Renal agenesis** is the absence of one or both kidneys. Bilateral renal agenesis is incompatible with life. Unilateral renal agenesis is usually asymptomatic and may go undiagnosed in the absence of other congenital anomalies. Renal agenesis affects about one in every 1,360 infants in North Carolina.

### Musculoskeletal Defects

Musculoskeletal defects are a heterogeneous group of conditions that involve the bones, cartilage, muscles, connective tissue, body wall, and diaphragm. Affecting about one percent of all live births, they are also one of the most common group of anomalies.

**Gastroschisis** is an opening in the abdominal wall, typically to the right of the umbilicus (the site where the umbilical cord joins the abdomen) (Figure 16a). The opening, which is usually less than two inches, allows the abdominal viscera to protrude through the body wall and into the amniotic sac. Surgical correction is required. About one in every 2,820 infants is affected in North Carolina. For reasons that are not clear, gastroschisis is much more common among infants of very young mothers and, in recent years, the prevalence has been increasing in North Carolina as well as in other areas.

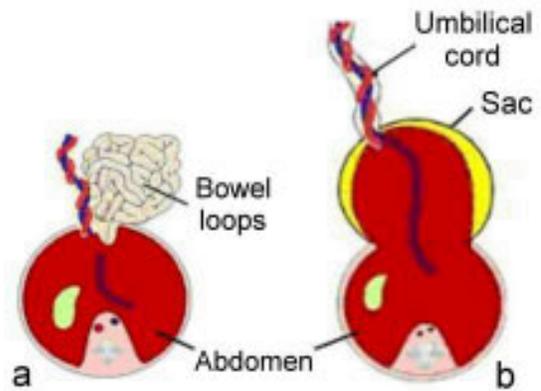


Figure 16. Gastroschisis (a); Omphalocele (b) (cross-sectional views)

**Omphalocele** is a hole in the abdominal wall ranging in size from less than an inch to an area covering most of the abdomen (Figure 16b). Parts of the intestinal tract, covered by a membrane, protrude through the hole at the umbilicus. The size of the defect usually correlates with the amount of intestinal tissue involved. When the opening is large, the entire intestinal tract, including the stomach, liver, and spleen, may be affected. Omphalocele is often associated with other anomalies, including chromosomal disorders. Surgical correction is required to repair omphalocele, and prognosis is good if the infant has no other serious congenital malformations. Omphalocele affects about one in every 5,600 infants in North Carolina.

### Chromosomal Disorders

Chromosomes are the inherited, microscopic structures that house an individual's hereditary information in the form of genes. Humans normally have 23 pairs of chromosomes (46 total) in each cell of their body. Chromosomal anomalies typically arise from an abnormal number of chromosomes or from certain defects in specific segments of the chromosomes. These conditions can occur spontaneously or can be inherited. Examples of chromosomal disorders include trisomy 21 (Down syndrome), trisomy 13 (Patau syndrome), Klinefelter syndrome, and Turner syndrome. Chromosomal disorders can cause structural (physical) birth defects, mental retardation, fetal and infant death, and shortened life expectancy.

One of the more frequently occurring chromosomal disorders is trisomy 21, which is commonly referred to as **Down syndrome**. Individuals with Down syndrome have an extra whole or partial chromosome 21 (Figure 17). Some degree of mental retardation is typically present, along with physical features including epicanthal folds, upward slanting eyes, large tongue, broad and short hands, a single crease in the palm (simian crease), and cardiac abnormalities. First trimester spontaneous abortion occurs in over half of affected pregnancies. About 40 percent of infants with Down syndrome have cardiac anomalies, the majority of these being atrioventricular septal defects. Survival after birth is

influenced by the presence or absence of the associated cardiac abnormalities. About one in every 750 infants born in North Carolina has Down syndrome. Women ages 35 and above are at a much higher risk for having pregnancies affected by Down syndrome and other trisomies as compared to younger women. The reason for the increased risk among older women is not well understood.

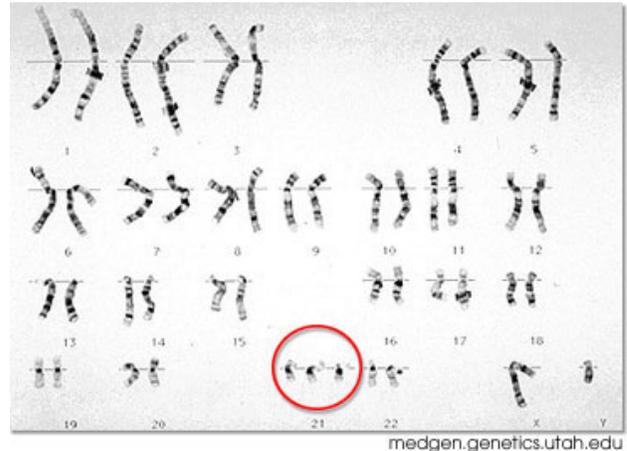


Figure 17. Karyotype of infant with Down syndrome (note extra copy of chromosome 21)

## Birth Defects Prevention

By taking precautions before and during pregnancy, a woman can reduce her risk of delivering a baby born with a birth defect or other adverse outcome. It is very important that women start planning for the health of their baby before becoming pregnant.

During the first three to eight weeks after conception, many of the baby's vital organs and systems are being formed. By the time most women know they are pregnant, their baby's development is well underway, and some birth defects may have already occurred. While there is never a guarantee for a healthy baby, the following list of preventive measures can increase a woman's chance of having a healthy pregnancy and a healthy baby.

### **Talk with your health care provider**

Prior to pregnancy, it is a good idea to talk with a health care professional. During this time, a health care provider can identify any health risks a woman may be facing and work with her to address them before she becomes pregnant. It is important to have conditions such as diabetes, epilepsy, and high blood pressure under control before becoming pregnant. If there is a history of an inherited or genetic disorder, consultation with a genetic counselor may be recommended.

### **Consume folic acid**

Several studies have shown that women who take a daily multivitamin with 400 micrograms of folic acid before and during pregnancy decrease the risk that their baby will be born with a neural tube defect by up to 70 percent. Consuming folic acid may also prevent other birth defects, such as cleft lip/cleft palate and some congenital heart defects. For adults, folic acid may offer protection from illnesses such as heart disease and colon cancer.

### **Eat a healthy diet**

Women and their developing babies can benefit from good nutritional habits before and during pregnancy. All women should eat a well-balanced and varied diet and take a multivitamin every day.

### **Exercise regularly**

Regular exercise can benefit a woman's body by increasing overall strength and by creating a healthy environment in which her baby can develop. Talk

with a health care provider to determine an appropriate exercise level.

### **Maintain an ideal weight**

The preconceptional period is an excellent time to achieve and maintain an ideal weight. Women who start their pregnancies underweight or overweight may have problems. If a woman is overweight at the time of conception, she is more likely to develop high blood pressure and diabetes during pregnancy, and is also at increased risk for certain birth defects. If a woman is underweight, she is more likely to deliver a low-birth-weight baby.

### **Avoid smoking**

Women should avoid smoking during pregnancy, and limit exposure to secondhand smoke. Smoking during pregnancy is associated with an increased risk of miscarriage and stillbirth, sudden infant death syndrome (SIDS), and low birth weight. In addition, children exposed to smoke may have behavioral problems, learning difficulties, and an increased risk for respiratory problems and asthma.

### **Avoid alcohol**

The harmful effects of alcohol on a fetus' growth and development are numerous. Fetal alcohol syndrome (FAS) is the most severe, creating physical, mental, and behavioral problems in infants. Alcohol consumption during pregnancy is the leading cause of preventable mental retardation among infants.

### **Avoid illicit drugs**

Research has shown that in-utero exposure to illicit drugs can cause direct toxic effects on a developing fetus, as well as create fetal and maternal dependency. The baby may experience withdrawal prenatally when drugs are withdrawn from a dependent mother, or after delivery.

### **Limit exposure to environmental hazards**

Pregnant women should minimize exposure to toxic substances and chemicals. They should also avoid eating undercooked meat and handling cat litter, as these activities may lead to an infection known as toxoplasmosis, which can seriously harm a developing fetus. There are a few foods, including certain types of fish, some soft cheeses, and ready-to-eat meats, which may also pose a risk during pregnancy.

### **Discuss medications**

All medications – prescription or over-the-counter – that a woman may be taking should be discussed with a pharmacist or health care provider, as some may not be appropriate to use during pregnancy.

### **Check immunizations**

It is important for a woman to check her immunization history before pregnancy. If she is not immune to chickenpox and rubella, or has not received her hepatitis B series, she should talk with her health care provider about her risks.

Women with questions about any of the above should talk with their health care provider.

## **Genetic Services for Children and Families**

In recent years, the field of medical genetics has grown at an extremely rapid pace. Advancements in the field have created an increasing need for health professionals trained to explain genetic information to families. The North Carolina Genetics and Newborn Screening Unit, part of the Division of Public Health, provides comprehensive genetic services for any infant, child, adult, or pregnant woman suspected of having a genetic condition.

Genetics consultations or evaluations may be helpful for individuals who have:

- A genetic disorder (e.g., trisomy 21, Huntington disease);
- A family history of birth defects;
- Mental retardation or a family history of mental retardation;
- A racial or ethnic background with a higher incidence of certain disorders (e.g., Tay Sachs disease, sickle cell disease);
- A history of cancer, heart disease, or certain other diseases;
- Maternal age during pregnancy of 34 or older; or
- An abnormal ultrasound or maternal serum screening result during pregnancy.

During a genetic evaluation, a genetic counselor and/or clinical geneticist may:

- Gather information regarding the reason for referral;
- Review the medical, family, and pregnancy histories;
- Review medical records;
- Describe the diagnosis under consideration and issues regarding the diagnosis;
- Order testing for a disorder;
- Interpret the results of physical examinations and tests;
- Communicate the information to the family;
- Support the family and help with coping skills; and
- Follow up and maintain ongoing communication with the family.

## Appendix. Prevalence\* of selected birth defects, North Carolina, 2003

Birth Defect (BPA code)	Number of Cases	Prevalence	95% Confidence Interval
<b>Central Nervous System</b>			
Anencephaly/acrania (740.000 - 740.100)	29	2.45	1.64 - 3.52
Spina bifida w/o anencephaly (741.000 - 741.990)	60	5.07	3.87 - 6.53
Spina bifida with hydrocephalus (741.000 - 741.090)	35	2.96	2.06 - 4.12
Spina bifida w/o hydrocephalus (741.900 - 741.990)	25	2.11	1.37 - 3.12
Encephalocele (742.000 - 742.090)	11	0.93	0.46 - 1.66
Microcephalus (742.100)	88	7.44	5.97 - 9.17
Holoprosencephaly (742.260)	7	0.59	0.24 - 1.22
Hydrocephalus w/o spina bifida (742.300 - 742.390)	136	11.50	9.65 - 13.60
<b>Eye/Ear</b>			
Anophthalmia/microphthalmia (743.000 - 743.100)	20	1.69	1.03 - 2.61
Congenital glaucoma (743.200)	9	0.76	0.35 - 1.44
Congenital cataract (743.320 - 743.326)	26	2.20	1.44 - 3.22
Aniridia (743.420)	2	0.17	0.02 - 0.61
Anotia/microtia (744.010; 744.210)	21	1.78	1.10 - 2.71
<b>Cardiovascular</b>			
Common truncus (745.000 - 745.010)	13	1.10	0.06 - 1.88
Transposition of great vessels (745.100 - 745.190)	63	5.33	4.09 - 6.81
Tetralogy of Fallot (745.200 - 745.210; 746.840)	44	3.72	2.70 - 4.99
Single ventricle (745.300)	28	2.37	1.57 - 3.42
Ventricular septal defect (745.400 - 745.490)	515	43.54	39.86 - 47.46
Atrial septal defect (745.510 - 745.590)	506	42.78	39.14 - 46.66
Endocardial cushion defect (745.600 - 745.690)	67	5.66	4.39 - 7.19
Pulmonary valve stenosis/atresia (746.000 - 746.010)	112	9.47	7.80 - 11.39
Tricuspid valve stenosis/atresia (746.100)	16	1.35	0.77 - 2.20
Ebstein's anomaly (746.200)	10	0.85	0.41 - 1.56
Aortic valve stenosis/atresia (746.300)	31	2.62	1.78 - 3.72
Hypoplastic left heart syndrome (746.700)	26	2.20	1.44 - 3.22
Patent ductus arteriosus** (747.000)	571	48.27	44.40 - 52.39
Coarctation of aorta (747.100 - 747.190)	69	5.83	5.54 - 7.38
Total/partial anomalous venous return (747.420 - 747.430)	27	2.28	1.50 - 3.32
<b>Respiratory</b>			
Choanal atresia (748.000)	23	1.94	1.23 - 2.92
Lung agenesis/hypoplasia** (748.500 - 748.510)	15	1.27	0.71 - 2.09
<b>Orofacial</b>			
Cleft palate w/o cleft lip (749.000 - 749.090)	65	5.49	4.24 - 7.00
Cleft lip with or w/o cleft palate (749.100 - 749.290)	111	9.38	7.72 - 11.30

\*Number of cases per 10,000 live births.

\*\* Excludes infants < 2,500 grams.

## Appendix. Prevalence\* of selected birth defects, North Carolina, 2003 (cont.)

Birth Defect (BPA code)	Number of Cases	Prevalence	95% Confidence Interval
<b>Gastrointestinal</b>			
Tracheoesophageal fistula/esophageal atresia (750.300 - 750.380)	36	3.04	2.13 - 4.21
Congenital hypertrophic pyloric stenosis (750.510)	246	20.80	18.28 - 23.56
Stenosis/atresia of small intestine (751.100 - 751.195)	61	5.16	3.95 - 6.62
Stenosis/atresia of large intestine, rectum, anus (751.200 - 751.240)	65	5.49	4.24 - 7.00
Malrotation of intestines (751.400 - 751.495)	56	4.73	3.58 - 6.15
Hirschsprung's disease (congenital megacolon) (751.300 - 751.340)	36	3.04	2.13 - 4.21
Biliary atresia (751.650)	13	1.10	0.59 - 1.88
<b>Genitourinary</b>			
Renal agenesis (753.000 - 753.010)	87	7.35	5.89 - 9.07
Bladder exstrophy (753.500)	7	0.59	0.24 - 1.22
Obstructive genitourinary defects (753.200 - 753.290; 753.600 - 753.690)	273	23.08	20.42 - 25.98
Hypospadias/epispadias (752.600 - 752.620; 752.625 - 752.627)	398	33.65	30.43 - 37.11
<b>Musculoskeletal</b>			
Congenital hip dislocation (754.300)	94	7.95	6.42 - 9.72
Club foot w/o CNS defect (754.500; 754.730)	66	5.58	4.32 - 7.10
Reduction defect of upper limb (755.200 - 755.290)	38	3.21	2.27 - 4.41
Reduction defect of lower limb (755.300 - 755.390)	25	2.11	1.37 - 3.12
Craniosynostosis (756.000 - 756.030)	52	4.40	3.28 - 5.76
Diaphragmatic hernia (756.610 - 756.617)	34	2.87	1.99 - 4.02
Gastroschisis (756.710)	42	3.55	2.56 - 4.80
Omphalocele (756.700)	21	1.78	1.10 - 2.71
<b>Chromosomal</b>			
Trisomy 21 (Down syndrome) (758.000 - 758.090)	157	13.27	11.28 - 15.52
Trisomy 13 (758.100 - 758.190)	11	0.93	0.46 - 1.67
Trisomy 18 (758.200 - 758.290)	36	3.04	2.13 - 4.21
<b>Other</b>			
Heterotaxia/situs (759.300 - 759.390)	16	1.35	0.77 - 2.20
Pierre Robin sequence (524.080)	13	1.10	0.59 - 1.88
Amniotic band sequence (658.800)	14	1.18	0.65 - 1.99

\*Number of cases per 10,000 live births.

\*\* Excludes infants < 2,500 grams.

## Resources

State Center for Health Statistics  
[www.schs.state.nc.us/SCHS](http://www.schs.state.nc.us/SCHS)  
919-733-4728

North Carolina Division of Public Health  
[www.ncpublichealth.com](http://www.ncpublichealth.com)  
919-733-7081

March of Dimes  
[www.modimes.org](http://www.modimes.org)  
1-888-MODIMES

North Carolina Folic Acid Council  
[www.getfolic.com](http://www.getfolic.com)  
1-800-367-2229

Spina Bifida Association of North Carolina  
[www.sbanc.org](http://www.sbanc.org)  
1-800-84-SBANC

The Cleft Palate Foundation  
[www.cleftline.org](http://www.cleftline.org)  
1-800-24-CLEFT

National Down Syndrome Society  
[www.ndss.org](http://www.ndss.org)  
1-800-221-4602

Family Support Network of North Carolina  
[www.fsnn.org](http://www.fsnn.org)  
1-800-852-0042

North Carolina Family Health Resource Line  
1-800-367-2229

National Birth Defects Prevention Network  
[www.nbdpn.org](http://www.nbdpn.org)

National Center on Birth Defects and Developmental Disabilities, CDC  
[www.cdc.gov](http://www.cdc.gov)

## Figure Credits

**Figure 1-3:** Courtesy of Dr. Roger Stevenson, Greenwood Genetics Center, Greenwood, South Carolina.

**Figure 4-10:** Manitoba Pediatric Cardiac Surgery Inquest Report, Manitoba Provincial Court, Winnipeg, Manitoba.

**Figure 11-12:** Used with kind permission of the Cleft Palate Foundation, Chapel Hill, North Carolina ([www.cleftline.org](http://www.cleftline.org)).

**Figure 13:** Tracheo-Oesophageal Fistula Support (TOFS), Nottingham, UK. Used with permission.

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**Figure 17:** Department of Genetics, University of Utah, Salt Lake City, UT.

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