



national • birth • defects • prevention • study

2015-2016

NEWS

NBDPS News is distributed from the Centers for Disease Control and Prevention (CDC)-funded Centers for Birth Defects Research and Prevention.

The purpose of this newsletter is to inform study participants and the public of the Centers' activities and current news about birth defects.

NBDPS Next Steps

Thank you for participating in the National Birth Defects Prevention Study (NBDPS). As NBDPS interviewing has come to a close, we will continue to analyze NBDPS data for years to come. We are excited to have launched a new study to continue the effort to learn more about the causes of birth defects. This new study is called the Birth Defects Study To Evaluate Pregnancy exposureS, or BD-STEPS. BD-STEPS furthers the work of other studies, like the NBDPS, that have been going on for more than 25 years.

About BD-STEPS

BD-STEPS began conducting phone interviews with women living in seven states—Arkansas, California, Georgia, Iowa, Massachusetts, New York, and North Carolina. BD-STEPS participants include women who have had a pregnancy affected by a birth defect as well as mothers of babies who do not have a birth defect.

The BD-STEPS interview focuses on a variety of topics. A main area of focus is modifiable risk factors, which are risk factors for birth defects that might be changed to reduce the chance of having a child born with a birth defect. For example, BD-STEPS interviewers ask women detailed questions about medicines they take for longstanding diseases like asthma and high blood pressure. We know many women must take medicine to treat these diseases. However, a woman may be able to take a different medicine with a lower risk for causing birth defects. In this case, taking a different medicine is an example of a modifiable risk factor that she can change before and during pregnancy. BD-STEPS hopes to learn which medicines could pose fewer risks for birth defects.

BD-STEPS builds on knowledge from previous studies like NBDPS. NBDPS researchers identified some birth defect risks that need more research. For instance, NBDPS researchers learned that obesity and diabetes can increase a woman's risk of having a child with certain types of birth defects. With BD-STEPS, researchers will further explore obesity and diabetes, and focus on behaviors women can change. This new research will give women more information about how they can reduce their risks for having a child with a birth defect.

Currently, BD-STEPS primarily uses phone interviews to collect information. However, BD-STEPS is looking for better and easier ways for women to take part in the study. Some women may be asked to complete BD-STEPS surveys online to help us develop new ways for doing birth defects research.

Learning about birth defects takes time and involves many people. We learn something from every interview, but there is still more work to do. BD-STEPS is part of a strong and continued commitment by researchers to learn even more about the causes of birth defects.

In this



- NBDPS Next Steps ----- 1
- Recent Findings from the NBDPS ----- 2
- Exploring the Relationship Between Medication and Birth Defects ----- 5
- Spotlight on NBDPS Research Centers and the Principal Investigators ----- 6
- Parent's Viewpoint ----- 10
- Babies to Adults: Special Note to Study Participants ----- 10
- Resource Corner ----- 11
- Directory of the Research Centers ----- 12

Recent Findings from the NBDPS

NBDPS is no longer interviewing participants. Over the course of 14 years of interviews, 43,000 women from 10 states took part in the study. We also have cheek cell kits (genetic samples) from more than 25,000 families that will be used to understand how genetics play a role in birth defects.

Although NBDPS data collection is complete, we will continue to analyze this rich source of information for years to come. More than 200 scientific and medical papers using NBDPS data have already been published. Here are some of the recent research findings.

Polycyclic Aromatic Hydrocarbons and Oral Clefts

Polycyclic aromatic hydrocarbons (PAHs) are chemicals formed during the burning of coal, oil, gas, garbage, or from smoking cigarettes, cigars, or pipes, or from charbroiling meat. There are many ways to come in contact with PAHs at home or at work. NBDPS researchers looked for evidence of a link between PAHs in a woman's workplace and oral clefts ([clefts of the lip and palate](#)) in their babies.



Among women interviewed for the NBDPS, researchers found that it was not very common for women to work in places with PAHs. PAH exposure occurred most often among women working in food preparation and serving. Pregnant women who worked in places with any PAHs were more likely to have babies with cleft lip with or without a cleft palate. Pregnant women who worked in places with higher PAH levels were even more likely to have babies with cleft lip than women who worked in areas with lower PAH levels. Since this was the first study to show this link, more research is needed to understand it fully.

Langlois PH, Hoyt AT, Lupo PJ, et al. Maternal occupational exposure to polycyclic aromatic hydrocarbons and risk of oral cleft-affected pregnancies. *The Cleft Palate-Craniofacial Journal*. 2013;50(3): 337-46.

Butalbital Use and Birth Defects

Butalbital (byoo-TAL-bi-tal) is an ingredient in some medicines (for example, Fioricet®, Fiorinal®) prescribed for the treatment of migraine and tension-type headaches. Little is known about the safety or risk of using butalbital while pregnant. NBDPS researchers looked to see if there is a link between butalbital use and birth defects.

Researchers found that butalbital use is rare among women in NBDPS. Researchers also found a link between using butalbital while pregnant and certain [congenital heart defects](#). The heart defects in the study with this link include tetralogy of Fallot, pulmonary valve stenosis, and atrial septal defects. This study is the first to show these links. More research is needed to learn more about the risks and benefits of butalbital use among pregnant women.



Browne ML, Van Zutphen AR, Botto LD, et al. Maternal butalbital use and selected defects in the National Birth Defects Prevention Study. *Headache*. 2014;54(1):54-66.

Antiherpetic Medication and Gastroschisis

[Gastroschisis](#) is a birth defect of the abdominal (belly) wall. The baby's intestines are outside of the baby's body through a hole beside the belly button. The causes of gastroschisis are not known, but it is thought that exposures in early pregnancy might play a key role.

Herpes infections can occur during pregnancy, and to treat these infections antiherpetic medicines are sometimes used. Researchers used NBDPS data to see if antiherpetic medicine use in early pregnancy is linked to the risk of gastroschisis. This study found that the risk of gastroschisis could be higher in children born to women with herpes infection who used antiherpetic medicine.



More research is needed to determine the risks of herpes and antihherpetic medications in pregnancy.

Ahrens KA, Anderka MT, Feldkamp ML, et al. Antihherpetic medication use and the risk of gastroschisis: findings from the National Birth Defects Prevention Study, 1997-2007. *Pediatric and Perinatal Epidemiology*. 2013 Jul; 27:340-5.

Genetics and Congenital Heart Defects

Genetics and [congenital heart defects](#) (CHDs) are the most common type of birth defect, affecting about one in every 100 babies born in the United States. Scientists are working hard to learn the causes of CHDs. Most CHDs are due to a complex relationship among environment, lifestyle, and genetic factors.



Using the genetic samples that NBDPS families provided, NBDPS researchers identified changes (sometimes called variants) in several genes that were linked with CHDs. Variants in four genes were found to increase the risk of heart defects. More research is needed to understand the functions of these genes and to confirm the study findings among different groups. This study is one of the first in NBDPS to study genes from babies and their mothers.

Li M, Cleves MA, Mallick H, et al. A genetic association study detects haplotypes associated with obstructive heart defects. *Human Genetics*. Sep 2014;133(9):1127-38.

Fever in Pregnancy and Congenital Heart Defects

Fever in early pregnancy is common. It is reported by 5% to 10% of women during the first trimester. [Congenital heart defects](#) (CHDs) are the most common birth defect. NBDPS researchers looked to see if there is a link between fever during pregnancy and CHDs.

Among mothers of a baby without a birth defect, one out of 13



had a fever in early pregnancy. For mothers of a baby with a CHD, about one in 12 had a fever in early pregnancy. Researchers found that the risk for CHDs was higher if the fever was from a urinary tract infection or pelvic inflammatory disease than if it was from a respiratory, flu-like disease. The increased risk was seen for certain CHD types, such as heterotaxy and some obstructive heart defects. Results of this study suggest the risk for these heart defects might be lower in women who were taking multivitamins before their pregnancy.

Botto LD, Panichello JD, Browne ML, et al. Congenital heart defects after maternal fever. *American Journal of Obstetrics and Gynecology*. 2014 Apr;210(4):359.e1-359.e11.

Air Pollution and Congenital Heart Defects

Many pregnant women, especially those who live in urban areas, are exposed to different air pollutants. Although the Environmental Protection Agency sets air quality standards to limit the amount of pollution in our air, we still do not know whether the amounts of pollution women are exposed to can affect their unborn babies.

NBDPS researchers found that higher exposure to air pollutants related to car traffic, specifically nitrogen dioxide, was more common among mothers of babies born with certain types of [congenital heart defects](#) (CHDs). More research is needed to learn what levels of air pollution affect an unborn baby.

Stingone JA, Luben TJ, Daniels JL, et al. Maternal exposure to criteria air pollutants and congenital heart defects in offspring: results from the National Birth Defects Prevention Study. *Environmental Health Perspectives*. 2014 Aug;122(8):863-72.



The Potential Impact of Newborn Pulse Oximetry Screening

In the United States, about 7,200 babies born each year have [critical congenital heart disease](#), also known as CCHD. CCHDs are the most serious types of CHDs. CCHDs often lead to low levels of oxygen in a newborn. A low oxygen level in a baby can be found by testing oxygen levels 24 hours after birth.

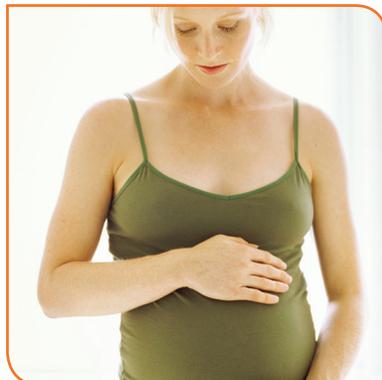


NBDPS researchers found that about 30% of the babies with CCHDs were diagnosed more than three days after birth. The percentage of babies with more than three days delay in diagnosis varied based on CCHD type—ranging from about 8% for babies with pulmonary atresia to 62% for babies with coarctation of the aorta. If these babies had been tested for CCHD within 24–48 hours of birth, as is now mandated in many state newborn screening programs, some of them might have been diagnosed sooner, leading to earlier treatment.

Peterson C, Ailes E, Riehle-Colarusso T, et al. Late detection of critical congenital heart disease among US infants: estimation of the potential impact of proposed universal screening using pulse oximetry. *Journal of the American Medical Association (JAMA) Pediatrics*. 2014;168(4):361-70.

Risk Factors for Choanal Atresia in the NBDPS

Choanal atresia is a narrowing of the nasal airway and can cause severe breathing problems in newborns. It is one of the leading causes of nasal surgery. Choanal atresia is rare, and not much is known about its risk factors. NBDPS researchers looked for reasons that might increase the risk for having a baby with choanal atresia.



Researchers found that the risk of choanal atresia was higher when women smoked cigarettes, had three or more cups of coffee with caffeine every day, or used thyroid medications early in their pregnancy. This study is one of the first to look at risk factors for choanal atresia, so more research is needed to confirm these findings.

Kancherla V, Romitti PA, Sun L, et al. Descriptive and risk factor analysis for choanal atresia: the National Birth Defects Prevention Study, 1997–2007. *European Journal of Medical Genetics*. 2014;57(5):220-9.

Participation in the Genetic Component of NBDPS

Most women (63%) who finished an NBDPS phone interview also gave NBDPS genetic samples by providing swabs of their cheek cells. NBDPS researchers found that taking part in the genetics part of the study differed by Center. The participation ranged from 50% to 74% across different sites.

Participation also differed by race/ethnicity. Among white and Hispanic women, those with a higher education and had a baby with a birth defect were more likely to complete the cheek cell collection than women of other races. Among all racial/ethnic groups, an extra \$20 incentive and a shorter time between the baby's due date and the phone interview were linked with women being more likely to participate. Researchers can use this feedback to increase participation in similar studies in the future.

Glidewell J, Reefhuis J, Rasmussen S, et al. Factors affecting maternal participation in the genetic component of the National Birth Defects Prevention Study—United States, 1997–2007. *Genetics in Medicine*. 2014 Apr;16(4):329-37.



Exploring the Relationship Between Medication and Birth Defects

Many women take medicines during pregnancy. In fact, we have learned from the NBDPS and other studies that nearly 90% of women in the United States take one or more medicines during their pregnancies, including a large number who take prescription medicines. Even more women take medicines that are available without a prescription, called “over-the-counter”, or OTC, products. Reasons for taking medicine range from treating longstanding illnesses such as diabetes, high blood pressure, and asthma, to treating more short-term problems such as a urinary tract infection or a cold.

With so many women taking medicines while pregnant, it is very important to learn as much as we can about whether these products might harm (or help) a pregnancy. We know now that some medicines may cause birth defects if a woman takes them while pregnant. For example, thalidomide, isotretinoin (Accutane), and methotrexate may harm the developing baby. At the same time, we have learned that some vitamins can reduce the risk of certain birth defects. For example, [folic acid](#) has been shown to reduce the risk of severe birth defects of the spine and brain. However, for most medicines, we know very little about their possible effects when taken during pregnancy. Women and their healthcare providers need to know which medicines to try to avoid and which are the safest to take during pregnancy.

Our new study, BD-STEPS, studies medicines by asking questions during the interview about any medicine use during the first trimester of pregnancy. Our focus is on the first trimester because it is when most birth defects occur. We ask women to tell us about all the medicines they took, including prescription and OTC products, vitamins, supplements, and herbals. We want to know about pills and liquids as well as shots, suppositories, and inhaled medicines. Then we ask women to tell us when during pregnancy they took these products and how often they took them. We ask women to try to report as accurately as they can by looking at calendars and prescription bottles.

Because most birth defects are quite rare, we must gather data from many women. We will combine the medications information already obtained from the NBDPS interviews with the new BD-STEPS medications information; this will greatly strengthen the birth defects data. Once we have studied these data carefully, BD-STEPS researchers will be able to help women and their healthcare providers make better choices about the medicines women decide to take in pregnancy.



Spotlight on NBDPS Research Centers and the Principal Investigators

Did you know that all of the NBDPS Research Centers work together to continue analyzing data, but each Center is unique? Many of these Centers will continue their work in researching birth defects in BD-STEPS. Read below for more information about the NBDPS Centers and the Principal Investigators at these Centers.

ARKANSAS

Each year in Arkansas, around 1,300 babies are born with a birth defect and more than 100 babies will die because of them. The Arkansas Center for Birth Defects Research and Prevention seeks to reduce the occurrence of birth defects in Arkansas and the nation. The Center collects its data through the Arkansas Reproductive Health Monitoring System (ARHMS), one of the oldest active birth defects surveillance systems.

Research at the Arkansas Center looks at genetic factors that might increase the risk for birth defects. Its participation in NBDPS and BD-STEPS provides the opportunity to look more closely at genes, a woman's lifestyle, and the interaction between genes and exposures in the woman's surroundings that might lead to birth defects. The goal of the research is to find exposures in early pregnancy that may be prevented.

The Arkansas Center's Principal Investigator is **Charlotte Hobbs, MD, PhD**. Since 1997, Dr. Hobbs has been the PI for the Arkansas Center (during NBDPS and now in BD-STEPS) and the Medical Director of ARHMS. Dr. Hobbs oversees study analyses, presents findings at meetings and in scientific journals, and takes a lead role in mentoring new birth defects researchers. Dr. Hobbs has devoted her career to the study and prevention of birth defects.



and Prevention since 1997. The California Center focuses its research on the impact of nutrition, environmental exposures, and genetics on risks of birth defects.

Gary Shaw, DrPH, and Suzan Carmichael, PhD, are the Principal Investigators for the California Center. Dr. Shaw has conducted birth defects research for over 25 years. Dr. Carmichael has focused on birth defects research for the past 15 years. Together, Drs. Shaw and Carmichael are leaders in birth defects research and have written hundreds of scientific papers on the topic. They have researched causes related to diet, obesity, drugs, alcohol, stress, pollution, jobs, and genes.



GEORGIA (CDC)

CDC coordinates NBDPS data-management and analyses and BD-STEPS study activities and serves as the Georgia study site. CDC brings a lot of experience, like studying medication use among pregnant women and finding new ways to look at the data. In addition, CDC keeps track of birth defects in Atlanta through the Metropolitan Atlanta Congenital Defects Program (MACDP). MACDP has collected data in Atlanta since 1968 and serves as a model for other state birth defects tracking systems.

More than 3,000 women from Georgia have helped us understand the causes of birth defects by taking part in the NBDPS. The Georgia Center is eager to follow up on NBDPS research findings with BD-STEPS.

The Principal Investigator for the Georgia Center is **Sarah Tinker, PhD**. Dr. Tinker focuses on collecting high-quality data from local study subjects. She works with those involved in the



Georgia site to ensure that the study is running smoothly. She also analyzes data with a focus on using novel statistical methods.

The overall lead investigator at the CDC for the collaborative NBDPS and BD-STEPS centers is **Jennita Reefhuis, PhD**. She continues to work with a team of computer programmers, communication specialists, and scientists to coordinate the overall study logistics for BD-STEPS as she did for NBDPS. Dr. Reefhuis' research interests include fertility treatments and medications, and how they relate to birth defects.



IOWA

The NBDPS and BD-STEPS study areas in Iowa include the entire state of more than 3 million people. Iowa has a mix of urban and rural residents, each displaying differences in personal behaviors, such as smoking and alcohol use. In addition, some Iowans are exposed to chemicals from farms and other toxins in the environment. Iowa researchers started the Iowa Registry for Congenital and Inherited Disorders (IRCID) partly to study how these exposures might affect birth defects. Every year, the IRCID monitors more than 40,000 live births and fetal deaths in Iowa, and each year over 1,500 pregnancies are affected by birth defects.

The Iowa Center has expertise in studying personal behaviors (smoking and alcohol use) habits, as well as chemicals and toxins, as risk factors for birth defects. The Iowa Center has also been a leader in studying genes that might play a role in birth defects. For NBDPS and BD-STEPS, the Iowa Center will look at the impact of personal behaviors and environmental exposures, along with genes, on birth defects.

Paul Romitti, PhD, leads the Iowa Center as the Principal Investigator. Dr. Romitti also leads the IRCID and has worked with the registry since 1989. Dr. Romitti's research focuses on environmental exposures and the role of genes in birth defects. He has also researched study methods and led a long-term follow-up of children with birth defects.



MASSACHUSETTS

The Massachusetts Center started in 1997 and is a partnership between the Massachusetts Department of Public Health, Boston University's Slone Epidemiology Center, and the Genetics Unit at MassGeneral Hospital for Children. The Massachusetts Center has experts in many research areas. These areas include public health tracking, health studies of children and pregnant women, and heart defects.

The Massachusetts Center is also a leader in studying the effects of medicine use during pregnancy. Massachusetts researchers showed that medicine use during pregnancy has gone up over the past 30 years. The effects of prescription and over-the-counter drugs and herbal supplements on babies is often not known. As part of NBDPS, the Massachusetts Center continues to study the effects of medicine use during pregnancy.

Marlene Anderka, ScD, MPH, is the Principal Investigator and the Director of the Massachusetts Center at the Massachusetts Department of Public Health since 2001. She directs the Center's NBDPS and BD-STEPS work and decides which research the Center plans. She works closely with two co-investigators: Allen Mitchell, MD, Director of the Slone Epidemiology Center at Boston University, and Lewis Holmes, MD, Director (emeritus) of the Genetics Unit at MassGeneral Hospital for Children.



NEW YORK

In New York State, there are about 250,000 births every year. Over 12,000 of these babies will have a major birth defect. The New York Center has experts who study medicine use during pregnancy, as well as environmental exposures at work and other places that might lead to birth defects. The Center often collaborates with the Wadsworth Center, the research-intensive public health laboratory at the New York State Department of Health. Their colleagues at Wadsworth have developed cutting-edge ways to do genetic and environmental analysis of newborn screening blood spots for birth defects research.

Charlotte Druschel, MD, MPH, and Marilyn Browne, PhD, served as the Principal Investigators for the New York Center for NBDPS.

Dr. Druschel was an integral part of the New York Center for 17 years and retired in 2014.

Dr. Druschel is a reproductive and perinatal epidemiologist who has more than 25 years of experience in birth defects research. Her most recent publications have focused on maternal illnesses and medications and identifying environmental and genetic risk factors for specific birth defects. Dr. Druschel has also played a key role in developing methodology for the tracking of fetal alcohol syndrome (FAS) and muscular dystrophy. Dr. Browne is the PI for BD-STEPS. Dr. Browne has worked in birth defects research for more than 15 years. Her focus is gaps in knowledge about the risks and benefits of different ways to treat a woman's longstanding medical conditions.



For BD-STEPS, as with NBDPS, Dr. Browne and her team hope to use their research to learn more about which medicines pregnant women can safely use to treat longstanding health problems. Dr. Browne has published studies on thyroid medication use during pregnancy and use of the migraine medicine, butalbital, as well as studies on caffeine use during pregnancy. She is also involved in tracking adolescents and adults with [congenital heart defects](#) to learn more about their health care needs, and studies of genetic risk factors for birth defects.



NORTH CAROLINA

Each year in North Carolina, more than 3,500 babies are born with major birth defects. Despite new findings, the causes of most birth defects are still unknown. The North Carolina Center carries out birth defects research in 33 counties of central North Carolina. Children with birth defects are identified through the North Carolina Birth Defects Monitoring Program (NCBDMP) that has been run by the state since 1995. The North Carolina Center has two partners: the Department of Epidemiology at the University of North Carolina (UNC) Gillings School of Global Public Health in Chapel Hill, and the NCBDMP at the State Center for Health Statistics, Division of Public Health, in Raleigh.

The North Carolina Center works to find exposures during early pregnancy that put women at higher risk of having a baby with a birth defect. This research may lead to public health efforts to prevent future birth defects. For example, the North Carolina Center has studied the impact of nutrition (including diet and obesity) and toxins in the air and drinking water on pregnancies in relation to birth defects. For BD-STEPS, as with NBDPS, the North Carolina researchers will focus on these topics:

- Risk factors for birth defects during pregnancy that might be modified, including diet, obesity, exercise, and work exposures;
- The role of genes and gene- environment factors; and
- New methods of studying birth defects.

Andrew F. Olshan, PhD, and Robert Meyer, PhD, lead the North Carolina Center as Co-Principal Investigators.

Dr. Olshan's research looks at how genes and the environment affect reproduction, birth defects, and cancer. He was among the first researchers to explore how a father's work might be a risk factor for birth defects. As its Principal Investigators, Dr. Olshan directs the North Carolina Center's research, staff, and study-related activities. Dr. Meyer directs the North Carolina Birth Defects Monitoring Program (NCBDMP). His primary role in the North Carolina Center is to oversee the clinical data collection activities of the Center and to participate in the design and conduct of local and pooled studies using NBDPS data. His current research interests focus on understanding potential environmental causes of birth defects and on long-term outcomes among children and families, including survival, educational achievement, and quality of life.



TEXAS

There are roughly 380,000 births each year in Texas. Over 20,000 of those will have a major birth defect recorded in the Texas Birth Defects Registry (TBDR). The TBDR has been operating since 1994, and is housed in the Texas Department of State Health Services. The Texas Center for Birth Defects Research and Prevention is composed of staff at the TBDR and partners across the state, including the Schools of Public Health and Medicine at the University of Texas, University of Texas (Austin), University of Texas Southwestern (Dallas), Texas State University (San Marcos), Texas A&M Public Policy Research Institute and School of Public Health (College Station), and Baylor College of Medicine (Houston).

Researchers at the Texas Center are interested in exploring:

- Why certain race/ethnic groups have higher risk of birth defects;
- What causes birth defects, like [anencephaly](#) and [spina bifida](#) (severe birth defects of the spine and brain);
- The interaction between genes and diet or environmental factors on birth defects;
- What happens to children with birth defects – how long they live, quality of life;
- Occupational and environmental risk factors for birth defects, such as chemicals that occur in the workplace or result from water disinfection; and
- Other chemicals as potential risk factors for birth defects, like nitrate or similar substances found in food, water, and certain medications.

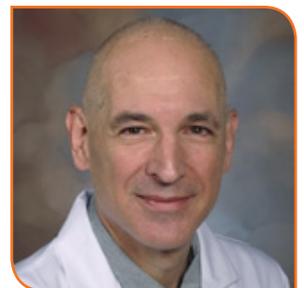
Mark A. Canfield, PhD, and **Peter Langlois, PhD,** have been with the TBDR since its first year and are co-Principal Investigators for the Texas Center for Research and Prevention.



UTAH

The Utah Center for Birth Defects Research and Prevention began in 2002 as a partnership between the Utah Birth Defect Network (UBDN) in the Utah Department of Health and the Department of Pediatrics at the University of Utah. The University of Washington joined the group in 2005. The Utah Center included women throughout the state from 2003 through 2011. Utah Center researchers have focused their efforts on improving the understanding of the environmental and genetic risk factors for [congenital heart defects](#), [gastroschisis](#), and [cleft lip and palate](#). Congenital heart defects are common and make up 25% of all birth defects in Utah. Gastroschisis, though not as common, has increased among all babies born over time for reasons that are still unclear. Cleft lip and palate rates in Utah are among the highest in the nation and the world. Finding the causes of these birth defects will help us understand ways to lower the risk of birth defects and help families have healthy babies.

Marcia Feldkamp, PhD, PA, MSPH, and **Lorenzo Botto, MD,** are co-Principal Investigators for the Utah Center. Dr. Feldkamp, an Associate Professor of Pediatrics at the University of Utah, is interested in the contribution of genetics, maternal infections and inflammation as risk factors for birth defects. Dr. Botto is a Professor of Pediatrics at the University of Utah and is a board certified pediatric geneticist and medical epidemiologist with a research emphasis on genetic and environmental factors that increase the risk for congenital heart defects.



Parent's Viewpoint

Our daughter, Shelby, was born in 2007 with an isolated soft cleft palate that was not detected at our 20-week ultrasound. The night she was born, they put her to my chest to nurse. After several frustrating attempts to get her to latch on, the nurse examined her further. She discovered that Shelby had a cleft palate. I had no idea what a cleft palate was. I had heard of a cleft lip but did not know you could be born without the roof of your mouth. After talking to the lactation specialist, I was really disappointed to learn I would not be able to nurse at all. They said for her, it would be like drinking out of a straw with a hole in it because she could not get proper suction. I chose to pump milk for her, which I would end up successfully doing for a whole year.



In addition to a cleft palate, Shelby also had a very small chin. It almost seemed like her face ended at her mouth, a condition called Pierre Robin Sequence. She would need to eat from special bottles with tops called Pigeon nipples. She would need to be held upright to eat. She would have to sleep on her stomach so that she would not aspirate (have liquid go down into her lungs) in her sleep and so her tongue would not block her airway. She needed to be hooked up to a monitor to make sure she was getting enough oxygen. Even though we were not prepared for all of these things before she was born, for us there was no other choice but to stay positive and informed. By the time we left the hospital two days later, we already had an appointment with a craniofacial surgeon (a surgeon who specializes in repairing birth defects of the head and face) at the children's hospital for the following week.

The first night home with Shelby was definitely the hardest. When she spit up, it would come out her nose. Sometimes an entire feeding's worth of milk would come out at once. I felt as if all I ever did was pump and feed her to keep her from starving and have extra for the times when she would spit it all back out.

At six months, she had her surgery to repair her palate. They said that she would not need any additional surgeries. It is

also common practice that when a cleft is repaired, tubes are placed in the child's eardrums to drain fluid that collects behind the eardrums that can cause hearing problems, so she had tubes put in. After her surgery, we were not sure what to expect for the future. What would her speech be like? Would her teeth come in normally?

She started to talk like any other kid would. In fact, her speech and vocabulary were higher than average! She has had three sets of ear tubes and unfortunately has not grown out of the need for them yet. Her teeth came in like they should, although her mouth was still very small, so we began her treatment with a pediatric orthodontist (a dentist who specializes in preventing and correcting crooked teeth). In the past few months, she began losing her baby teeth and the permanent ones coming in don't have very much room, so we're close to the next step in her orthodontic care. We're still not really sure how long the orthodontic process will be, but we know that it will be quite extensive (and expensive). As for the Pierre Robin Sequence, her jaw grew on its own and she did not need surgery to bring it forward. We had tests done and found that she did not have any other complications that may occur with Pierre Robin Sequence.



We have been very lucky to have excellent care and education along the way. It's so important for the staff in hospitals and doctors' offices to be informed and up-to-date so that they can give those in this situation the tools that they need. It's also really helpful to have a strong support system. A long time ago, this could have been a really scary situation. Today, this is something that is manageable. It doesn't have to be devastating. Shelby has come through every step of the journey thus far with flying colors! She's an energetic, stubborn, sassy, silly, intelligent seven-year-old. I feel so honored to share our story with others and I hope that other families can be inspired to be proactive for their children and know that there are resources out there to guide them through.

Editor's Note: This family has allowed to us to share their story. The privacy of study participants is important to us. We will not name anyone in the study in any report or publication unless we have been given permission to do so. Family stories are an important part of our newsletters and websites. We hope other families will share their stories with us.

Babies to Adults: Special Note to Study Participants

NBDPS has interviewed mothers for more than 16 years! Some babies of the earliest participants are close to reaching the age of 18. Participants can withdraw themselves and/or their child from the study at any time. In addition, when a child reaches the age of 18, he or she can also decide to withdraw from the study at any time. Withdrawing for a child means we will destroy the cheek-cell

sample and the data from the sample that we received when he/she was a baby. That means the sample will no longer be used. To withdraw from the study, call 404-498-4315 or e-mail NBDPS2@cdc.gov. For any person who withdraws from the study we will not use their data in the future, but we cannot remove their data from findings or articles that have already been published.

Resource Corner

Listed below are several resources that might be of interest. The Centers are not responsible for the content found on these websites.

Medication and Pregnancy

The American College of Allergy, Asthma, and Immunology has a webpage on asthma and allergies during pregnancy. It provides guidance for pregnant women who are dealing with these conditions.

http://www.acaai.org/allergist/liv_man/pregnancy/Pages/default.aspx

The **Mother To Baby** website contains a library of factsheets about different medications. These sheets answer questions about use of each medicine during pregnancy and breastfeeding.

<http://www.mothersbaby.org/fact-sheets-s13037>

CDC and partners are working together to provide better information to women and their healthcare providers about medication use during pregnancy. **Treating For Two**, an index site of CDC.gov, provides this information and other resources.

<http://www.cdc.gov/treatingfortwo/>

Stress and Pregnancy

The **March of Dimes** website on life changes during pregnancy talks about what causes stress and ways to reduce stress during pregnancy.

http://www.marchofdimes.com/pregnancy/lifechanges_indepth.html

The **Mother to Baby** website has a fact sheet on stress during pregnancy, available in English and Spanish. It covers what stress is, if moms-to-be should be concerned, ways to reduce stress, and where to go for help.

<http://www.mothersbaby.org/files/Stress.pdf>

Congenital Heart Defects

CDC's website has an entire index of webpages on **congenital heart defects**, including information on specific heart defects, research, and statistics, among other useful resources.

<http://www.cdc.gov/ncbddd/heartdefects/>

CDC's website also has a webpage on **congenital heart defects**. The webpage has information on screening, current research activities, and information for healthcare providers.

<http://www.cdc.gov/ncbddd/heartdefects/cchd-facts.html>

Cleft Lip and Palate

CDC's website has a webpage on **cleft lip and cleft palate**, providing information on what these conditions are, some of the known causes of cleft lip and palate, and diagnosis and treatment options.

<http://www.cdc.gov/ncbddd/birthdefects/cleftlip.html>

The Cleft Palate Foundation has information for parents of children with cleft lip with or without cleft palate.

<http://www.cleftline.org/parents-individuals/>

Children's Craniofacial Association website has information about birth defects of the head and face. They have resources on connecting with other parents and families as well as information on each condition.

<http://www.ccakids.com>

Choanal Atresia

The **Children's Choanal Atresia Foundation** has a website that provides information, research, and support about choanal atresia, or birth defects of the nasal passage.

<http://choanalatresia.org/index.html>

Gastroschisis

CDC's website has a webpage on **gastroschisis**. It talks about what gastroschisis is, how common it is, how it is diagnosed, and how it is treated.

<http://www.cdc.gov/ncbddd/birthdefects/gastroschisis.html>

Avery's Angels is a foundation that is helping children and families affected by gastroschisis. The website has resources for connecting with other families and ways to raise awareness about gastroschisis.

<http://www.averysangels.org/>

Genetics

CDC has a unique **Family Health History and Genetics** index of webpages that provide a great deal of information on the role genetics plays on the health of families, newborn screening, and many other useful resources.

<http://www.cdc.gov/ncbddd/genetics/>

Directory of the Research Centers

To reach a NBDPS or BD-STEPS study coordinator by phone, please call (404) 498-4315. Please see below for specific contact information to each BD-STEPS Center.

ARKANSAS

Charlotte Hobbs, MD, PhD

University of Arkansas for Medical Sciences
Arkansas Children's Hospital
E-mail: ar@bdsteps.org
<http://arbirthdefectsresearch.uams.edu>

CALIFORNIA

Suzan Carmichael, PhD

Gary Shaw, DrPH

Stanford University
E-mail: ca@bdsteps.org
<http://www.cdph.ca.gov/programs/cbdmp/Pages/default.aspx>

GEORGIA/CDC

Jennita Reefhuis, PhD

Sarah Tinker, PhD

Centers for Disease Control and Prevention
E-mail: ga@bdsteps.org
<http://www.cdc.gov/ncbddd>

IOWA

Paul Romitti, PhD

University of Iowa
E-mail: ia@bdsteps.org
<http://www.public-health.uiowa.edu/ircid>

MASSACHUSETTS

Marlene Anderka, ScD, MPH

Massachusetts Department of Public Health
E-mail: ma@bdsteps.org
<http://www.mass.gov/dph/birthdefects>

NEW YORK

Marilyn Browne, PhD

New York State Department of Health
E-mail: ny@bdsteps.org
http://www.health.ny.gov/diseases/congenital_malformations/

Note: Charlotte Druschel was a Principal Investigator for NBDPS but has since retired and cannot be reached through the NY Center.

NORTH CAROLINA

Andrew Olshan, PhD

University of North Carolina, Chapel Hill

Robert Meyer, PhD

North Carolina Department of Health & Human Services
E-mail: nc@bdsteps.org
<http://www.schs.state.nc.us/units/bdmp/>

TEXAS

Mark Canfield, PhD

Peter Langlois, PhD

Texas Department of State Health Services
E-mail: tx@nbdps.org
<http://www.dshs.state.tx.us/birthdefects/>

UTAH

Lorenzo Botto, MD

Marcia Feldkamp, PhD, PA, MSPH

Utah Department of Health
E-mail: ut@nbdps.org
<http://health.utah.gov/ubdn/>

share your stories

Newsletter Ideas and Mailing:

Please contact your Center listed in the directory if you:

- Want to share your experience about the NBDPS,
- No longer wish to receive this newsletter,
- Need to update your mailing address, or
- Would like to receive the newsletter via e-mail.

Also, please let us know if you have topic ideas for future issues.



Centers for Disease Control and Prevention
National Center on Birth Defects and Developmental Disabilities