

BD-STEPS News is a periodic newsletter of 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.

BD-STEPS Update

We are excited to have launched a new study to help us learn more about the causes of birth defects. This study is called the Birth Defects Study To Evaluate Pregnancy exposureS, or BD-STEPS. BD-STEPS continues the work of other studies, like the National Birth Defects Prevention Study, or NBDPS, that have been active 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 conditions 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.



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Spotlight on BD-STEPS Research Centers and the Principal Investigators

Seven Centers across the United States work together as part of BD-STEPS. Each site brings unique experience and expertise to the research efforts. Read below for more information about the seven BD-STEPS 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.



from women who live in eight counties in the Central Valley. The Center has been part of the CDC-funded Centers for Birth Defects Research 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.

CALIFORNIA

Fifteen percent of all U.S. births occur in California. Each year, 17,000 babies are born with birth defects in California, and as many as 2,000 of these babies will die in their first year of life. The California Center of Excellence is a partnership with Stanford University School of Medicine and the California Birth Defects Monitoring Program in the Department of Public Health. The Center collects data

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.

Marilyn Browne, PhD, is the Principal Investigator for the New York Center. 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 health conditions. 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.



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 PI, 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.



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.

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.

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

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CALIFORNIA

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<http://www.cdph.ca.gov/programs/cbdmp/Pages/default.aspx>

GEORGIA/CDC

Jennita Reefhuis, PhD

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IOWA

Paul Romitti, PhD

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<http://www.public-health.uiowa.edu/ircid>

MASSACHUSETTS

Marlene Anderka, ScD, MPH

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NEW YORK

Marilyn Browne, PhD

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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

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Robert Meyer, PhD

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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