

From Beginning to End, **A BETTER LIFE FOR ALL**



National Center on Birth Defects and Developmental Disabilities

A Healthy, Happy Life

It may be the greatest wish people have for themselves and their loved ones. But for the millions of Americans with birth defects, disabilities, and blood disorders, that wish can be challenging to realize.

Life brings all of us complications and sometimes these include health problems. Staff of the National Center on Birth Defects and Developmental Disabilities (NCBDDD) work to protect those who are vulnerable to health risks, such as babies, children, people with blood disorders, and people with disabilities. Our ultimate goal is to promote health and enhance the potential for full and productive lives throughout the lifespan.

EVERY DAY OUR SCIENTISTS ARE ENGAGED IN:

- Saving babies through birth defects research and prevention.
- Understanding autism to help children live to the fullest.
- Preventing and managing complications from blood disorders, such as blood clots and hemophilia.
- Making sure that people with disabilities are not left behind, that health services and programs work for each individual's entire lifespan so that every person counts.

Please visit **cdc.gov/ncbddd** to learn more about the Center.

Follow Center Director, Dr. Coleen Boyle, on Twitter at **twitter.com/drboylecdc** to receive live updates regarding the Center focus areas.



Saving Babies through Birth Defects Research & Prevention

One in every 33 babies in the United States is born with a birth defect. Birth defects are a leading cause of infant deaths. These are two powerful statements. Our work begins before a child is conceived and continues throughout life. We're using our research to find ways to prevent birth defects and to improve the health of babies born with them. From our research we know that a woman's health before, during, and between pregnancies can affect the health of her baby. That's why we created a wealth of information on issues like folic acid, diabetes, smoking, obesity, drinking alcohol, and medication use during pregnancy. CDC has a unique and critical role in the national effort to address birth defects through state-based tracking and public health research. It is through our collaborations with states, academic centers, and other partners that we turn this information into action that prevents birth defects and improves the lives of those affected.

Kristine's Story

After a healthy and normal pregnancy, I gave birth to my daughter, Cora, on November 30, 2009. Two days later we took her home, after getting a clean bill of health at the hospital. The next three days were spent cuddling and getting to know each other.

One morning my husband handed Cora to me because she seemed hungry. I started feeding her and looked up at my husband to tell him I loved him. When I looked back down, Cora was pale, gray, and not breathing. We raced to the hospital, which was no more than 5 minutes away, but it was too late. Cora was gone. The coroner told us that she died from an undetected congenital heart defect. Neither of us had heard of the term.

We later learned about a type of screening—pulse oximetry—that can help identify certain heart defects at birth. While we'll never know if the screening would have made a difference for Cora, we wish she'd had it. Cora's story is extremely sad, but it's also full of hope. In Indiana, where we live, a new law requiring the screening with pulse oximetry is named after her and is known as Cora's Law.

For now, I hope for a day when no mother finds out about her child's heart defect from a coroner. I hope that undetected congenital heart defects become a thing of the past. And ultimately, I hope that these birth defects can be prevented and that no baby is born with a broken heart.





Understanding Autism to Help Children Live to the Fullest

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Mary Elizabeth's Story

Our journey into this world of autism began eight years ago with the birth of our son, Perry. Since the beginning, he struggled. He seemed very restless. He had a lack of recognition in his eyes, didn't eat, and didn't sleep. As each day passed we recognized more 'red flag' characteristics of autism with repetitive behaviors and speech/sound delays.

There was a minimum six-month wait for him to see a developmental pediatrician. Meanwhile at 12 months old, we learned he was eligible for our state's early intervention program due to his developmental delays. He began a steady course of daily therapies. When Perry was 15 months old we finally received the diagnosis: Perry has autism. I cried for 2 days, hoping that I would be able to provide him everything he needed.

Shortly after Perry's diagnosis, our daughter Margeaux was born blue and immediately

faced feeding and sleeping problems. She was restless, but in a different way than Perry. When Margeaux was 12 months old we underwent another early intervention evaluation. Margeaux's therapy began and Perry's therapy continued. Her Asperger's diagnosis came at age 5. It has been a long road.

My incredibly special children have been a true gift. Like any mother, I would take away their struggles if I could. My ultimate hope is that my children will live in a world where they will be accepted and appreciated despite their differences. As long as I am here, I surround them with people who love and accept them because autism never takes a day off.





Improving the Lives of People with Disabilities

People with disabilities need health programs and services for the same reasons anyone does—to be well, active, and part of the community. Our work in disability and health focuses on sustaining lifelong health. We collaborate with national and state partners to ensure no one with disabilities is left behind; that every person counts.

Over 50 million people face greater health challenges and, compared to their peers without disabilities, are more likely to report poorer overall health, less access to adequate health care, and increased rates of smoking, physical inactivity, and chronic conditions. Disability—at birth or acquired through chronic disease, injury or aging—is part of the normal human experience. Yet people with disabilities are often overlooked in public health and health care practice.

It is vital that people with disabilities are included in health surveys, research, programs, policies and services that meet their needs as people, not just as people with disabilities. And when children and adults with disabilities receive needed programs and services across their lifespans they can reach their full potential, have an improved quality of life, and experience independence. We want to make every person count and are committed to equity in the health of people with disabilities at every stage of life.



Translating Science into Positive Change

Our research points us to effective strategies that help people with disabilities live healthier lives. We work with national and state partners to ensure no one with disabilities is left behind in our prevention efforts. Our research will answer important questions about the health care needs and quality of life of children and adults. We serve a critical role in identifying and communicating the needs of these populations.

Jerry's Story

I'm a 53-year-old father of four children. On December 3, 1976, I was hit by a drunk driver. The accident left me a partial paraplegic. But, my life is not defined by my disability. I live just like anyone else would. There is a lot I can do, and there are some things that I can't do.

I'm independent, have a house, raised a family, and my adult kids still look to me for support. I retired from computer programming in 2009, and compete in and coach several sports. I've even participated in the Boston Marathon.

As a person with a disability, however, I experience many barriers. Once, I was being prepared for surgery when a nurse proclaimed, "He doesn't need an epidural; he's a paraplegic." I had to inform the nurse that I was only a partial paraplegic and that I would, indeed, need an epidural.

I've seen a lot over 35 years of living with a disability. Many of the barriers and attitudes toward people with disabilities persist. But, there have also been many positive changes to get people with disabilities physically active through recreational opportunities, such as golf, fishing and even snow-skiing. There are now organizations such as Lakeshore Foundation—where I work part-time coaching youth basketball and track—that provide recreational opportunities.

I don't expect the world to revolve around me. I will adapt—just make it so I can adapt.



Understanding the Impact of Blood Disorders

Blood disorders affect millions of Americans, but we don't yet have an accurate picture of the real impact of these conditions. We do know that more than 20,000 people live with hemophilia. Sickle cell disease affects 90,000 to 100,000 Americans. As many as 1% of women in the United States may have a bleeding disorder and many are unaware of their condition. And we know that half a million or more Americans experience a new deep vein thrombosis (blood clot) each year; that at least 1 in 10 die—many without ever being diagnosed.

We evaluate multiple approaches for tracking the number of people who have blood disorders, like hemophilia and blood clots. This will help us target prevention efforts and identify where awareness of risk factors, signs, and symptoms needs to be raised. We know that it makes a difference when doctors and practitioners use health care recommendations, and that with early and accurate diagnosis and management, we can prevent and reduce the burden of blood disorders.

Blood disorders are a serious public health problem. CDC is uniquely positioned to reduce the public health burden resulting from these conditions by contributing to a better understanding of blood disorders and their complications. We ensure that prevention programs are developed, implemented, and evaluated, and that information is accessible to consumers and health care providers. We encourage action that improves the quality of life for people living with or affected by these conditions.



A black and white photograph of a woman with short dark hair, wearing a dark zip-up jacket over a light-colored shirt and dark leggings. She is running outdoors on a grassy path, holding a leash for a dark-colored dog whose tail is visible in the foreground. The image is partially obscured by a large, curved, light-colored graphic element on the left side of the page.

Mary's Story

My name is Mary Campise, and I would like to share my personal experience with blood clots, also known as deep vein thrombosis (DVT). I have been an avid runner and biker, and enjoyed good health my entire life. However, just before my 48th birthday, I began to experience shortness of breath. I found that I couldn't keep up running with my husband and friends. I began to struggle for breath just walking up stairs and finally decided to see a doctor. I was told that the shortness of breath could be the result of exercise-induced asthma or possibly an allergy. While I was trying to find an answer, my shortness of breath persisted.

Two weeks later I woke up to find that my left leg was twice the size of my right leg. My husband drove me straight to Texas Health Presbyterian Hospital of Dallas, where doctors told me that I had DVT. One thinks of a blood clot as being small. However, my sonogram showed a blockage that ran from my abdomen to my ankle, and I was told that no blood was moving up or going down. In the hospital I also learned that I had a complication of DVT—pulmonary embolism (PE). A PE occurs when the clot breaks off, travels to the heart and lodges in the lung. Most concerning to me, I learned that a PE can be fatal if not treated in time.

While I was in the hospital, a CT scan was conducted and the pulmonologist described my lungs as “being showered in blood clots.” An interventional radiologist talked with me that evening and explained that I needed a thrombectomy, an emergency procedure in which blood clots are surgically removed. After I was made aware of the risks and the necessity of this surgery, my husband and best friend convinced me that I needed to proceed in order to regain normal use of my leg. The very next morning I underwent a successful thrombectomy. I also had a special filter, called an inferior vena cava filter, inserted into that vein to prevent any more blood clots from moving into my lungs.

The best news for me was that when I came out of surgery my leg was once again normal in size and I could stand up on my legs with no problem! Doctors told me that it was very likely that my blood clot was caused by May-Thurner syndrome, a condition in which a vein on the left side is compressed by an artery on the right. I now take blood thinners, known also as anticoagulants, to manage my condition.

I am sharing my story because my experience with DVT is so similar to other people's stories—mainly that I did not recognize a warning sign, which for me was unexplained shortness of breath. My advice for other people is simple: be aware of your own body. If you experience a symptom such as unexplained shortness of breath, take this seriously and seek medical care immediately. Be your own advocate for your health and respond quickly to any symptoms that are not normal for you!

As our Work Continues, Hope Grows

By translating science into action, we provide the credible health information consumers, health care professionals, and policymakers need nationwide to improve the lives of children and adults with disabilities, blood disorders, and birth defects.

These statistics underscore the impact on individuals and families living with birth defects, disabilities, or blood disorders:

- One in five Americans has some type of disability.
- One in 33 babies in the U.S. is born with a birth defect.
- On average, 28,726 hospitalized adults with a blood clot diagnosis die each year.

At NCBDDD, we translate science into action to make a difference in the lives of these individuals, children, and adults. So as our work continues, there is good reason for hope to grow.

“ With more knowledge comes new hope for a healthier tomorrow for all. It’s rewarding to work in an environment that embodies this much passion for positive health impact. ”

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Centers for Disease Control and Prevention

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and Developmental Disabilities**

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