

Expanded newborn screening can save your child's life

Mary Lingle is a Web designer for Group M7 who lives in Tyler with her daughter Erin, 16.

By Mary Lingle

TYLER—Before she was born we worried about baby clothes and strollers and bassinets. We worried about which preschool she would attend, which high school and college. We worried about what it would be like if she got married and moved away from home—all this before we ever laid eyes on our beautiful daughter.

What we didn't know to worry about was expanded newborn screening.

So our only worries became which funeral home to call, which cemetery to bury her in, which headstone we should choose—and how to pick up the pieces of a broken heart.

Picture of health

I wrote the above after our daughter, Candice, died on Nov. 8, 1993, of a fatty-acid oxidation disorder called medium-chain acyl CoA dehydrogenase deficiency (MCAD). She was almost 3 years old and, on the morning of that tragic day, appeared to be the picture of health.

This so-called rare disorder with such a big name abruptly and with no warning took the life of our second child. One day she was playing with baby dolls and learning to do cartwheels with her big sister in our family room. The next day she was dead. Just like that.

The event that shattered us on the evening of Nov. 8 left us reeling and searching for answers. It took several weeks after the autopsy—which itself had taken two months—to get some of those answers.



Mary Lingle

The initial diagnosis was Reye's syndrome. Reye's syndrome? Wasn't that a rare brain and liver disorder we heard about in the '70s that was associated with the use of aspirin in children? How could this be? We didn't even have aspirin in our home.

It wasn't long before another finding came back. No, not Reye's syndrome but carnitine deficiency. The name sounded foreign and mysterious. Carnitine, a substance found naturally in meat, dairy products and breast milk, is crucial to many of the chemical reactions in the metabolic process. It works as a transport molecule, allowing cells to produce energy and remove toxic wastes. How could our child have carnitine deficiency with no outward signs?

Final diagnosis

Even though we were numb with grief, I had an insatiable desire to learn what had happened and why. I researched what I could but found little information in the early '90s about carnitine deficiency. I soon learned that carnitine deficiency, in our daughter's case, was secondary to the final, accurate diagnosis: MCAD.

Now we had a name. The information on MCAD was even more difficult to uncover than carnitine deficiency. I learned that it is autosomal recessive.

Autosomal means it is a condition that affects both boys and girls. Recessive means that both copies of the MCAD gene must be abnormal for a child to get the disease. Through DNA testing we learned that her father and I each contributed one defective gene. Her big sister also carries one defective gene. This was astounding. We carried this defective gene and passed it on to our precious child. Surely this must be a one-in-a-million chance? Not so.

One in 65 people in the population carries the defective MCAD gene. This is comparable to the one in 50 people who carry the defective phenylketonuria (PKU) gene. PKU?

I was familiar with PKU. Most people know about the heel stick all newborns in the United States receive shortly after birth that tests for PKU and a few other disorders. But what about MCAD? Why wasn't there a test that could detect MCAD and related metabolic disorders? What we learned shocked, saddened and infuriated us.

There was a test. In the late '80s the State of North Carolina was pioneering the way in expanded newborn screening. If our daughter had been born in North Carolina, chances are excellent that this deadly disorder would have been detected.

And what if it had been detected? What would have been the outcome for our daughter's quality of life? Would she die from MCAD? Would she be delayed mentally and physically? Again, what we learned led to more shock and anger.



BEFORE SHE WAS BORN—"What we didn't know to worry about before she was born was screening for childhood disorders that can kill," says Mary Lingle of her 2-year-old daughter Candice, shown here a few weeks before she died in 1993. [Courtesy photo]

10 or 12 hours to prevent blood-sugar levels from dropping too far. Children should eat high-carbohydrate meals when they are ill. A low-fat diet is recommended. Infants less than 1 year old should

those many years ago. We hear it all too often: "You mean there was a newborn-screening test for this disorder and we didn't know about it?"

So now I get to the real purpose of this article: expanded newborn screening.

There's an old saying: What you don't know can't hurt you. We learned the hard way that what you don't know can kill you.

The wheels of expanded newborn screening have turned excruciatingly slowly in this country. Look at a map of the states that offer expanded screening and you will see that the majority screen for fewer than 10 disorders. Currently 11 states screen for 30-plus disorders. It's encouraging to see progress, but for countless parents it's too little too late. Texas falls into the fewer-than-10 category.

We do have the occasional new parent who finds our group who comes to us with good news. Parents are so thankful that their state, or even their particular hospital that has a pilot program in place, has performed expanded screening that has resulted in an early diagnosis. Early diagnosis equals excellent results in their child's health. It can mean the difference between life and death.

What parents can do

What can parents do who live in states that lag in expanded newborn screening? For a fee of \$25 to under \$100, parents can pay out of their own pocket for the screening. For \$25 the Institute of Metabolic Disease at Baylor in Dallas offers expanded newborn screening for 30 additional disorders not included in state-required screening.

The Pediatrix Screening Lab in Bridgeville, Pa., will screen for more than 50 disorders for \$89. The Pediatrix Lab screening kit may even be purchased online and mailed to you or your child's doctor.

Moot point

I won't go into the controversies over whether laws implementing expanded newborn screening should or should not be mandated at the state level. The same arguments about accuracy, privacy, false positive tests and budget concerns have been in play for years. For those of us who have stood over that little white casket, the arguments are moot.

Our hope for all children is that they have an equal start in life.

Expanded newborn screening, although not perfect, is a tangible way for that to happen. Until every state in the United States screens for as many disorders as is technologically possible, our hope is to continue to spread awareness, to inform the masses.

We are the masses. I deeply regret that we weren't informed.

September is Newborn Screening Awareness Month. What you don't know about inherited disorders can kill your child.

MCAD and many disorders similar to MCAD are eminently treatable.

The main treatment is to avoid going without food for more than

continue to have at least one nighttime feeding or a late-night snack. Many people with MCAD take a prescription carnitine supplement to replace the carnitine the body cannot produce naturally.

That's it? Can this be true? What a small price to pay—if we'd only known.

And so the quest began. Through the Internet I was able to network with other parents and health-care professionals. I joined the Fatty Oxidation Disorders Support Group started by the parents of an MCAD child who died unexpectedly in 1985. I volunteered my time to produce and maintain the FODSupport.org Web site.

The support group, with the help of medical specialists and dietitians, focuses on disseminating information about fatty-oxidation and metabolic disorders. It also strongly advocates that all states implement expanded newborn screening.

What you don't know

Imagine that your child died in 1993 and 10 years later yet another family who has lost a child to one of these undiagnosed disorders finds you online and is just as shocked and angry as you were



THE COMPASSIONATE FRIENDS

The mission of The Compassionate Friends of Tyler is to assist families toward the positive resolution of grief following the death of a child of any age and to provide information to help others be supportive.

The Compassionate Friends is a national nonprofit, self-help support organization that offers friendship, understanding, and hope to bereaved parents, grandparents and siblings. There is no religious affiliation and there are no membership dues or fees.

The secret of TCF's success is simple: As seasoned grievers reach out to the newly bereaved, energy that has been directed inward begins to flow outward and both are helped to heal. The vision of The Compassionate Friends is that everyone who needs us will find us and everyone who finds us will be helped.

Our monthly group meetings are held on the third Tuesday of each month at the Clinical Associates of East Texas building at 2010 Sybil Lane (just off Paluxy Drive) in Tyler.

For more information, please call (903) 581-2831, (903) 780-7104 or visit us online at www.TylerTCF.org.



We need not walk alone.