Public Comments to the Advisory Committee on Heritable Disorders and Genetic Disease in Newborns and Children

Micki Gartzke, parent
Shorewood, WI

Mom of LeA Marie Gartzke (1996-1998)
Director of Education and Awareness
Hunter's Hope Foundation

June 8, 2004

Thank you ladies and gentlemen of the Committee for the opportunity to share my comments with you on this very important issue of newborn screening.

We are all here together at this time, this very special first time, to improve a process—Newborn screening—that overall we all know has not kept pace with the current needs of public health. I want you to know that I am here to help you help all children receive equitably distributed newborn screening and thus, their right to a healthy start in life.

As we all know, babies are dying unnecessarily because of lack of early identification and access to effective treatment. My daughter died because she did not receive early identification.

There is treatment for her disease, but it needs to be started within the first 60 days of life. My daughter did not get diagnosed until she was 10 months old, a full six months after we started looking for a diagnosis. Consequently, treatment was well out of the range of possibilities by then. I have a crystal clear picture of the importance of newborn screening. I know a little boy born with the same disease ten days before my daughter, he was identified at birth, my daughter at ten months. Today he is in the first grade and my daughter has been buried in her grave for five and 1/2 years. My daughter's medical bills have been calculated to be nearly a quarter million dollars in her two short years. My daughter's death is but one example of the need for universal newborn screening. This committee can greatly help many families in the United States by making recommendations to Congress about how to improve newborn screening. I need to share a few of the details our lives: my husband's, my daughter LeA's, and mine, so you can see inside of a family who has lost a child due to lack of early identification and all that that means. Loss of love, loss of companionship, loss of hopes, loss of dreams, loss of potential . . . just to mention a few.

Having married later in life, having had a long, productive and fulfilling career, I was more than excited to transition after our marriage and become a mom, a full-time stay at home mom. It seemed like a dream come true. Having been single well into my 30's I had seen a fair share of the world, I had family and friends, fulfilling work, a bountiful amount of love. But none of that prepared me for what I would encounter after my daughter was born.

On October, 14, 1996, her predicted due date, weighing in at 7.2 lbs and 19 3/4 inches, with a mop top head full of dark hair and wide-open big dark eyes, our daughter, LeA, burst into our lives and showered us with a new type of love. Her Apgars were 8's and 9's. Right from the beginning we assured her, "Someday you'll grow into your eyes, they were so big!" She was all eyes . . . . My mom said she took after me . . . I don't know. From the minute LeA arrived she was alert, looking around taking everything in. She was calm and peaceful with the typical little voice of baby cries, cries that were easily remedied by gentle touch or nourishment. Oh, I had been warned about the possible challenges of beginning breast-feeding. But LeA, she got it right away, no problem! What we didn't know at that time was that LeA was busy taking in her environment right away, as I suspect her soul knew that her time on this earth was not to be long. The first time my best friend met LeA she said, "LeA looks like an old soul, she looks wise, her gaze is deep, and it is almost like you can already hear her thinking behind those big dark eyes." And being proud new parents we felt as if we could.

Oh, what a honeymoon we all had when we brought our baby home from the hospital. She slept, she ate, and she looked around just like a new baby is supposed to. And her first smile, well the three of us melted

into one when that happened. For the first four months, we had such a delightful time with our " Baby LeA" as she was quickly becoming called. Me, a mom of advanced maternal age, boy I was right on top of everything. I noted LeA's milestones, and she hit them in the first few months right on cue. I was so proud. I can see her now sitting up in the corner of the couch, shaking a jiggly toy and laughing at the sounds that were coming out of it—and she, of course, had no idea why the sounds were being made. But they intrigued her, and she showed her pleasure with them. I can still see her sitting in her bouncy chair playing with the toys on the overhead toy bar, occasionally catching herself in the mirror and smiling at herself, almost as if flirting. I remember playing “peek-a-boo" in the mirror with the baby. We did that many times and Lea giggled so much during that game, life was wonderful. But those laughs and smiles and giggles were all soon to go away, once the crying started.

Being an on top of it kind of mom I scheduled LeA's four-month check-up when she was four months old exactly, on February 14, 1997. Our pediatrician examined her and declared that she was doing wonderfully and by the way, could he take her home for the weekend as he and his wife were really missing having little ones around. I assured him I understood his enthusiasm, as the past four months had been the most amazingly joy-filled time in my 37 years. After the doctor was done with his duties, and before I left that check-up, I pulled out my list of particulars that I wanted to ask and share with him—stuff like, when do I start with solids, how to introduce a bottle—you know the usual kind of stuff. The last item on my list turned out to be rather unusual, however, at the time I had no idea that what I had noted would have devastating consequences. I said, "So what's up with the thumbs. They used to be out," and I showed him what I meant, "and now they are just doing this tucked in thing." Again I showed him what I was beginning to see more and more on a regular basis, although not yet on a continuous basis. I looked up from my daughter's hands only to see my beloved pediatrician's face, and before my eyes, he turned white, the color drained from his face, and in the next split second, he said to me, "that thumb thing you described is called 'cortical thumbs' and it's generally indicative of a neurological problem. We'll have to get you all over to see some specialists and find out what's going on your little beauty queen." I felt like someone had just hit my head with a hammer. The full impact of that day did not hit us right away, as LeA continued to eat, grow, smile, giggle, and hit more milestones.

Shortly thereafter, we found ourselves standing right on a fault line at the epicenter of an earthquake. Our foundations were rocked. Uncontrollable, around-the-clock, unsoothable crying started. In one day, just like that (a snap of the fingers) we went from having a virtual lovefest with our daughter, to being cast as the three central characters in a dark and grim nightmare—so unimaginable by those who have not stood in similar shoes, that any description I could give would not bear the full weight of the darkness.

From happy, smiling and laughing, LeA bee-lined straight to crying—her body rigid, stiff as board. Not able to eat and inconsolable. DEVASTATING! It was an instantaneous change, as if someone had thrown a switch. The next six months were spent in what seemed like every possible clinical specialty at the Children's Hospital Clinics. From one misdiagnosis to the next, they had us orbiting the hospital, chasing hopes that something conclusive would come up. None of the doctors were looking for a zebra in their backyard, even though it was there all the time! Reflux, colic, cerebral palsy, we heard all the "umbrella" terms.

"We're sorry Mrs. Gartzke, her MRS shows that infant myelination is within normal ranges for her age," I was told. "What is myelination? How do we know it is normal? Can I see the pictures? Can you explain it?" I asked. I was looked at and treated as a mother who was overly concerned. They tried to soothe me instead of answering my questions.

IRATE does not begin to fully capture the mood that surrounded me. The very medical professionals that my daughter's care was entrusted to were not coming up with answers as I was being held hostage watching my daughter lose weight, eat slower, choke every time she tried to eat. With a feeding rate of an ounce an hour, she was choking. I called and shared these concerns with doctors. Again I got, " . . . oh Mrs. Gartzke. Sure, we'll make an appointment to get LeA into a feeding specialist, do a swallow study for starters." I can't tell you how many times I heard, "Mrs. Gartzke, the earliest appointment we have is 6 weeks from now." And in the meantime my darling baby was starving, and crying uncontrollably.

Through trial and error, we figured out that white noise and motion helped to offset a wee bit of the crying. Let me tell you, I feel as if I did not sit down for six months. We walked with LeA, she swung in her little

swing so much it burnt out a motor. But it made her happy. We drove LeA. We found a route with no stops. We could drive for an hour each direction, LeA did not cry. WE WERE EXHAUSTED. We worked so hard to soothe her that we had exhausted ourselves—and still we had no diagnosis! Finally, another MRS was ordered, still outpatient, and before I could get the results I was watching my daughter shrinking rapidly. She had been up to 17 lbs and she was down to 11.5. I felt as if I was watching her die in my arms. ENOUGH! I called the pediatrician whom I dearly loved and said, "Dear sweet doctor I have a dilemma, I believe LeA is dying in my arms today from starvation and no one is helping her find the exact problem, and I cannot take it at home anymore with her. I am leaving in one hour for the hospital and you have two choices, either I bring her through the ER (which I don't imagine will reflect well on you) or you get her admitted ASAP and call me back. I am leaving in one hour." He called me back. We were admitted. After the 20-minute car ride to the hospital LeA had perked up again and I even have a picture of her smiling in the waiting room at the hospital. One of her last smiles!

A seven-day stay in the hospital accomplished what had been unobtainable in the previous six months. And then the long-awaited impressions from the second MRS. I could tell by the doctors' attempt to not have long faces, that the news WAS NOT GOOD! "Mrs. Gartzke," that's all I let them say before I asked, "What are the impressions this time and please don't tell me inconclusive again. This child is dying, There is something wrong." We think it is a white matter problem and most likely a Leukodystrophy, specifically, Globoid Cell. Suddenly they were speaking Greek, or I was deaf. I wasn't sure, but none of it made sense! They still did not want to deal with the possible depth of the situation, insisting that we wait til the blood tests come back with conclusive results before they would talk to me about anything.

CRAZY, this is all CRAZY, I thought! I immediately called my Internet savvy friend and had her looking for anything and everything on globoid cell leukodystrophy. And within an hour, she was in my room with information from something called PUB MED with a list of citations of research projects done on this disease, as well as a Web site about a little boy named CJ, who also had been recently diagnosed with the same disease. Three days later, at the end of the day, the neurologist came in with a plethora of followers and confirmed one of the impressions from the MRS. LeA was deficient in an enzyme called galactocerebrosidase and this was causing her problems. By then I has learned about this disease through my own reading knowing that this enzyme was crucial to forming myelin during the vital early stages of brain development, and at best what he told me was minimal. "It's fatal, there's nothing you can do, take her home and make her comfortable. Call the ULF, they may be able to offer some parental support." Shortly thereafter he walked out of the room, his team of followers on his heals. In front of 15 strangers, in a small hospital room, the man had dropped a bomb on our family and he did not seem to find it out of the ordinary to convey such disastrous news to our family with an audience gawking at us. It felt like torture upon torture. Fatal, and your daughter, are words that should not be allowed to be used in the same sentence, especially when your daughter is smiling at you and she is ten months old. Well, that was the end of Stage Two, August 7, 1997. My birthday.

Stage Three started when we went home with an NG tube in my daughter, who was gaining weight like there was no tomorrow, she went directly from the 3-6 mos. size she had been wearing for six months to 18 month is like a week. The crying was abated by the use of nutrition and a couple of starter meds. LeA was scheduled for a feeding G-tube within weeks. A hospice nurse came to our house, the first of many, to teach us how to use the feeding pumps, tubes, suction machine, oxygen. By then we had already learned about CPT and were well ensconced in daily sessions of OT and PT. She remained stable for four months—if you can call feeding tubes, oxygen and suction machines stable? I'm not sure. New Year's Day LeA crashed. Her hospice worker came over and told us that she believed this was LeA's last day of life. Well, I tell you, after hearing," It's fatal, there's nothing that can be done," I didn't think I would ever hear more devastating news, but hearing, "I think this is Lea's last day of life," knocked that earlier statement right outta the ring.

That brings us to the last stage, Stage Four, for the remaining ten months of my daughter's life. This stage was full of feeding tubes, specialty formulas, suctioning, deep suctioning, I can do deep suctioning. A mother should not know how to deep suction her child. Pneumonias. 36 doses of medicine daily. DNRs. Learning all about how to fill out a DNR on your 13 month-old child is not something a parent should do. With the help of many hours of private duty nurses, and a great set of family and friends, we were able to

spend as much quality time as possible with our daughter as the final stages of her disease robbed her of life.

This story provides testimony to the significance of early detection. I will remind you again what I said earlier . . . I know a little boy born with the same disease ten days before my daughter. He was identified at birth, my daughter at ten months. Today he is in first grade and my daughter has been buried in her grave for five and 1/2 years.

Please help save other families from having to write stories like this.

The Children's Health Act of 2000 had promised to help fund states expanded newborn screening, but has yet to follow through with the money. States are left to their own means and I have heard that only 18 have decided that children's lives are worth the effort and cost.

I ask that this Committee encourage Congress to require states to inform parents in writing of the potential for their children to receive additional newborn screening tests that may not be required under State law. We must start by mandating that the hospitals educate parents on the availability of supplemental Newborn screening tests. This parental notification must be meaningful and require informed consent. New Jersey has recently enacted this type of legislation. We must immediately put a plan in place for adding all testable/treatable diseases to every state in the nation's Mandatory Newborn Screening list.

The solution seems so simple. Available today are screening tests, technology, and treatments they are all available today, right now. We need to use them. We need to fix our current NBS system so that currently available resources are used to give every child the right to a healthy life.