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Special Report — PART THREE

NEWBORN SCREENING

Newborn Screening and Parent Support Groups

by Kathy Stagni

Could it have been different? We will always wonder if life would have been different had newborn screening been available 11 years ago when my daughter Melissa was born with propionic acidemia (PA). Melissa was born on November 11, 1988, with an acute case of PA, an inborn error of metabolism that is so rare, the incidence rate is only about 1 in 50,000. Her metabolic doctor told us we would probably have better odds of hitting the jackpot in Las Vegas.

Even if the latest technology for screening newborns at birth, tandem mass spectrometry, had been available then, most likely it would not have saved Melissa from her initial episode. The onset of PA caused a coma, high ammonia levels in her blood, and brain swelling, resulting in mild mental retardation, speech delays, and the need for continuous feeding of a special low-protein formula through a nasogastric tube.

We will never know if Melissa’s outcome could have been prevented, but we believed we could make a difference for others. My mission for the last three years has been to help families connect and provide mutual support through a newsletter and the Internet. Three years ago, I took over the Organic Acidemia Association (OAA). Started by Lorie Asten-Neilson, OAA is a small, grassroots parent support group that has been in existence since the early 80s. OAA represents more than fifteen different organic acidemias, which involve the body’s inability to break down protein.

“WE ARE NOT ALONE”
I had heard of OAA from my daughter’s metabolic doctor shortly after her diagnosis, but out of denial, I did not contact them immediately. When I did, the OAA newsletter became the only outlet I had to share my feelings and experiences and learn about other families dealing with the same situation. Through the newsletter we even met a few families, some living in Minnesota as we do. That sense of connecting is very important in raising a child with a metabolic disorder. Parents have the knowledge and experience

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to help other families deal with the day-to-day problems that can occur while caring for a child with such life-threatening disorders. We have experienced so many similarities among these children that the sense of relief we feel in knowing we are not alone is one of the few emotional supports we have in dealing with such a rare disorder. I will never know the heartache of losing a child because of misdiagnosis, but I do know the heartache of raising a child with special needs and always wondering "what if?"

When the Internet began to get popular, I started to communicate via e-mail with a family living in Massachusetts who has a daughter with PA. I thought it was wonderful to be able to share ideas and stories with others who could relate, and we both thought it would be great to share our stories more widely on the Internet. The previous director of OAA was eager for my assistance, and I suddenly found myself as the executive director of the group. We started a Web site and began hearing from parents all over the world who had children with rare metabolic disorders.

OAA is starting to see positive benefits from expanded newborn screening. Just over a year ago, when their son, Mubashir, was born, the Younis family lived in Massachusetts, a state that has implemented expanded screening that includes organic acidemia disorders. The very day that the family learned that Mubashir was diagnosed with PA, they contacted the OAA over the Internet. Waheed Younis claims that Mubashir is doing better than any other PA kid that he knows about, something he credits to early intervention and expanded newborn screening! Waheed also praises the OAA's mission. "The OAA gave me lots of emotional support," he said. "Knowing that we are not alone makes it easier to accept the challenges. And we've also received lots of useful information that helps us manage Mubashir each and every day."

JORDAN FRANKS

Other families are not as fortunate. Jill Franks lives in Chicago, Illinois, where newborn screening has not been expanded. This past February, when her son Jordan was just 4-days-old, he had a metabolic crisis. Jill remembers bringing Jordan to the emergency room, where the doctors struggled to reach a diagnosis. Jordan's ammonia level went to 550 (10 times the normal range), which can lead to permanent brain damage. He was initially diagnosed with methylmalonic acidemia, and a day later the diagnosis was revised to PA when the lab results were completed. Jill often wonders if newborn screening would have identified the disease before Jordan's metabolic crisis. Jill says, "Words cannot express how much the support from OAA parents has meant to me and my family, including my mother. I have never encountered such an amazing group of positive people..."
NEWBORN SCREENING

Mubashir Younis, 1, was diagnosed with PA very early, thanks to the expanded newborn screening in Massachusetts. His parents credit Mubashir’s health today to early detection.

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ple. Every time I read the OAA newsletter, I relive the first days of Jordan’s life. As the saying goes, the names have changed but the story remains the same. OAA parents are more than new friends, they are like new family members. They gave me, and continue to give me, invaluable information on getting therapy early and often, what to expect, reminders that “it will get better,” and treatment plans. I only hope that I can be as helpful to new parents facing a diagnosis of PA.”

LISTENING TO THE STORIES

Cay Welch and her husband Mike Metil started a separate support group, International Organization for Glutaric Acidemia (IOGA), when their son Michael was born six years ago. Their group focuses on Michael’s specific organic acidemia, glutaric acidemia type I, a disorder that has a high incidence rate in the Amish community. Cay tells a wonderful story about meeting Carla, a 17-year-old Amish girl. When they met, Carla sat almost proudly in her wheelchair, although, Cay said, “She had to be all of 95 pounds—soaking wet.” Able to communicate only by moving her headband pointer, Carla fired off question after question. She told Cay her story of self-diagnosis, which began in the library and grew in chat rooms on the Internet discussing rare metabolic disorders. Soon after matching her symptoms with those typical of GA, she had an explanation for her mysterious headaches, muscle weakness, and nausea when digesting meat. Carla asked her mother to contact a clinic in Pennsylvania that she had found during her research, and she and her mother then traveled from New York State to Strasburg, Pennsylvania, to see Dr. Holmes Morton at The Clinic for Special Children. Carla’s diagnosis also gave the family answers to why Carla’s baby brother, Johnnie, was not sitting, crawling, or talking at 18 months old. Then there was the unexplained death of a sibling which was diagnosed as sudden infant death syndrome (SIDS). Did Johnnie and the baby with SIDS have GA? Newborn screening could have easily detected GA in this entire family and offered an opportunity for early intervention.

Cay tells another story, too, about Barbie Ann and her sister Lydia. In their matching blue and black pinafore dresses, they looked almost like twins, giggling, playing, and running barefoot through the yard where the Amish children bounded about. Shy but curious, they spoke some English and asked Cay what was wrong with Michael. “Why does he eat from a tube in his belly?” they asked. “Doesn’t he drink the same milk we do?” Cay answered, “Yes, but Michael had much brain damage when he was 11 months old and did not fare as well as you did.” The moment of realization that they had prospered, not suffered, despite their rare disorder was powerful for the Amish girls. And all because an expanded screen was made available at birth: a home birth, no less, which is common among Amish women. Cay adds, “Although I cannot rescue my son or wave a magic wand saying, ‘This will all be better,’ Michael, I can and do aid others through our IOGA Web site, newsletters, co-sponsoring medical conferences, linking families, and advocating the expanded newborn screening for every state.”

Organic acidemia disorders are highly manageable with diet and medication. Children who inherit these diseases are able to lead happy and productive lives. Newborn screening can improve their lives through early detection and intervention. It must be mandated across the United States as quickly as possible. Yes, my life is different from families with “normal” children, but I do feel my life has brought on a new meaning by having a child with a metabolic disorder. As Emra Bombeck wrote in her poem, “God chooses moms of special needs children for a purpose.” My purpose is to give Melissa the best quality of life she can have and to help support and connect the families that live with children with metabolic disorders.

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