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Area mother elected chair for national disease foundation

By Avani Nadkarni

Five years ago, Gene and Karen Quandt received some devastating news: Their only son, Ty, then 8 years old, was diagnosed with Niemann-Pick disease, a rare genetic disorder that affects just 500 people in the United States and that can cause dementia and be fatal.

After the initial shock, Karen Quandt, a registered nurse, decided to find out as much information as she could and became involved with the National Niemann-Pick Disease Foundation, based out of Milwaukee, Wis. In January, Karen Quandt was chosen as the chair of the foundation's board, a 12-person committee comprised of parents and family members of those with Niemann-Pick disease.

"(Karen) has brought just a wide array of talent and expertise, not only as a mom and a parent of a child with Niemann-Pick disease...but also as a trained nurse," Foundation Director of Family Services Nadine Hill said.

"She's a wonderful organizer, she's a taskmaster. She sees a problem and she can help make a determination as to what we need to do to address the situation and then it gets done."

Due to the rarity of the disease, Hill said, there is no government funding to fund a cure and there are no big pharmaceutical companies shelling out money, simply because there just isn't a huge patient base. So, Hill added, Karen Quandt and the foundation's work is more crucial to those affected.

Karen Quandt talks about the disease, her family and why she wanted to get involved:

Q: What is Niemann-Pick disease?

Karen Quandt: "It's a rare genetic disease. It's autosomal recessive, which means he has a chromosome with a mutation on it and he has a mutation from both parents. It causes problems for the body...it affects speech and swallowing and cognitive skills. It can affect any age group (but) mostly children are diagnosed. It's neurodegenerative, so it's progressive."

Q: What was your reaction when Ty was first diagnosed?

KQ: "I'd never heard of it so I tried to find out as much information as possible. You go to the Internet and you start looking things up. Someone had told me about the (National Niemann-Pick) Foundation so I did go to their Web site, too. You try to find people who have some sort of answers for you."

Q: How did you become involved with the foundation?

KQ: "We contacted the foundation about the time Ty was diagnosed and got a lot of information from them. We got involved and have been ever since. We had board meetings and in January, I was elected as board chair. It was pretty thrilling."

Q: What are your responsibilities now?

KQ: We have four board meetings a year, we get all the members on a call twice and we have face-to-face meetings in Wisconsin. We have one meeting with all the families (of people affected by the disease) and researchers with all the information. We have...to get involved with fundraisers. The majority of money comes from families...there is no treatment or cure right now, so we're looking to find the answers. There are less than 500 people diagnosed with the disease in the United States but it is very hard to diagnose so there could be more people affected. It's a close-knit group."

Q: What's been the best experience you've had on the board and as chair?

KQ: "Because I was (chair-elect), I was invited to an international meeting in October. There are many Niemann-Pick groups throughout the world and we had a meeting so we can talk about what we're doing and work as a group rather than many single groups. I met people from all over the world that are dealing with this disease. You don't feel like you're the only person...dealing with this...so that helps."

Q: Have you met other families in Washington dealing with Niemann-Pick disease?

KQ: Jim Lambright, (former) coach of the (University of Washington) Huskies. His two sons have the disease. I met his older son (and) we did get together. They are the only other people in the state of Washington, that I'm aware of."

Q: How is Ty now?

KQ: "It's been five years. He's on an experimental medicine and it is helping. It slows down the progression of the disease. He was having trouble swallowing and he was choking on foods...the medicine stopped that. It did slow the progression. He is able to go to school, he's a seventh-grader at Kalles Jr. High. He's able to speak, he can tell us what he wants and what he wants to do. He does have dementia, which makes school hard because they teach him something and it's hard for him to remember what it is. He can run and walk and play (but) fine motor skills are harder. He has a hard time holding a pencil. But he is able to do most things."