Monica Coenraads

MOTHER OF INVENTION

Paul Fetters

The birth of a first child is always a momentous event, as it was for Monica Coenraads when her daughter Chelsea arrived in October 1996. By Chelsea's first birthday, however, it was clear she was not developing normally. Six months later, Chelsea was diagnosed with Rett syndrome. But there was no definitive test for Rett syndrome, no known cause, and no treatments. Coenraads dedicated herself to seek out and work with scientists willing to study the disease. A cofounder of the Rett Syndrome Research Foundation (RSRF) and its current director of research, Coenraads has helped provide more than $11 million in research grants.

After Chelsea's diagnosis, I started to learn all I could about Rett syndrome. It was a pretty depressing scenario: The girls lost most if not all acquired language and motor skills, became autistic-like, and suffered seizures; some appeared to be in terrible pain. Even more depressing, the search for the gene had been ongoing for 15 years.

It was evident that an organization able to focus its energy on research would be beneficial. At the beginning, I just wanted to link scientists to speed discovery. I talked to top-notch scientists and directors of research organizations about what was needed. As it turned out, just as we were getting RSRF off the ground, the Rett-related gene *MECP2* was discovered, so we immediately began brainstorming about the next steps.

I never expected to fall in love with the science, but the neurobiology of Rett is fascinating. *MECP2* is involved in regulating the expression of other genes, so the question is which genes are responsible for the diverse neurological symptoms. *MECP2* has lured all sorts of researchers into the Rett field, because it touches on so many different areas of science.

Before the Foundation, I was running a successful restaurant. I have an MBA, but I really fell into the business. My father had opened a restaurant in Stamford, Connecticut, and soon after became very ill. My brother and I took over and ran it for 10 years. I figured that when life had thrown me lemons I had made lemonade once. When Rett syndrome reared its ugly head, I thought perhaps I could do it again.

I'm not so naïve to think that we have the resources to single-handedly cure Rett syndrome–that's bigger than any one organization, lab, or investigator. But we do have passion. Founded by six concerned parents of children with Rett, our organization is extremely motivated and a lot of what we do doesn't involve huge amounts of money. We can bring researchers together to share ideas. It's about engaging world-class talent. It's about forging relationships and pooling resources.

We can be nimble in ways that large federal agencies aren't, funding risky projects that the government isn't willing to take on. We have an esteemed scientific advisory board, chaired by Adrian Bird of the University of Edinburgh, and many ad hoc advisors who ensure that we fund high-impact projects directly relevant to Rett syndrome.

RSRF funding has played a part in nearly every breakthrough that's happened in the field since the *MECP2* gene mutations were discovered. For instance, we funded the development of an animal model of Rett that has proved crucial. Currently, we're funding collaborative work between HHMI investigators Huda Zoghbi and Nathaniel Heintz, targeting genes whose expression is up- or down-regulated because of *MECP2* dysfunction.

My style is very hands-on. I comb through scientific journals and press releases on a daily basis and regularly recognize work that could be synergistic with Rett syndrome. When I contact these scientists and engage them in discussion, they are intrigued by the scientific challenge of Rett.

In working with these researchers, I have definitely developed personal relationships. I think the fact that I'm a mother with a child with Rett personalizes it. Recently, we've created opportunities for researchers to meet our children, which I think helps them understand the urgency of the disease. It's true that science takes time and that good science takes even more time, but as a mother of a child with this syndrome, I know that we don't have time. And I think they get that.

During the last 7 years I've witnessed Rett syndrome rise from obscurity to become a high-profile disorder with links to autism, schizophrenia, and a host of other neurological diseases. It is my hope that Rett syndrome will prove to be the first treatable childhood neurological disorder.

INTERVIEW BY ALICIA AULT. *Monica Coenraads, her husband, Pieter, daughter Chelsea, and sons Alex, age 8, and Tyler, age 6, live in Trumbull, Connecticut.*