Code Talkers

Genome readers honor genetic counselors
who interpret complexity with compassion.

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INVITAE

Karlla W. Brigatti

A Gift for the Girls

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Nominated by
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Karlla Brigatti by Beth Buccini

My name is Beth Buccini, and I am the mother of a special needs daughter named Virginia. She was born in New York City in 2005 and was developmentally delayed from the beginning. For years, we relentlessly searched for answers — at Weill Cornell Medicine, NYU, and Mount Sinai Hospital, in New York; Mass General in Boston; Children's Hospital of Philadelphia; and A. I. DuPont in Wilmington, Delaware.

Finally, through a very personal connection, we ended up at an Amish and Mennonite clinic in the cornfields of Pennsylvania. At our first appointment at the Clinic for Special Children, after three hours with Karlla Brigatti and the head of the clinic, Dr. Kevin Strauss, they assured me that they could find an answer.

After visiting every major hospital on the East Coast for the past 10 years, I knew this place was very different. Never had I witnessed such compassion and dedication in an initial appointment. Three hours alone with both of them and my daughter! I felt I had stepped back in time to the way medicine must have been practiced decades ago. However, when Dr. Strauss and Karlla explained their scientific approach, I felt that I had sped forward into a different century.

Six months later, Karlla called me to say, "We know what your daughter has, and we know of only one other case in the world. Virginia has a de novo mutation of the DDX3X gene, which causes intellectual disability in girls."

This remarkable clinic discovers about five to 10 new gene mutations a year, and we happened to be one of them. By the end of the week, Karlla called me back to say, "We are up to seven cases." She located a group in the Netherlands that was also studying this gene and helped to author a paper published in the August 2015 edition of the *American Journal of Human Genetics*. The study included 38 girls with this disease.

Last August, just after the paper was published, she planned our first Family Day at the Clinic for Special Children, which nine of the 23 U.S. families known to be affected by this disorder attended. One and a half years later, she helped plan a second Family Day in Chicago, which 23 of the now 56 known U.S. families attended.

Karlla has continually dedicated much time and compassion to our nascent group and is a constant source of information, support, and networking. She is a tireless advocate for all of us, as if we are her full-time job. She has coordinated countless conference calls, explained basic genetics to parents over and over, steered us in the right direction for advocacy and fundraising, and answered many hard questions.

> **Her dedication, support, and compassion are unparalleled in an age when medicine has become so corporate. She is always willing to take a phone call, answer an email, or make a connection — literally even from a Brazilian rainforest.**

Thanks to Karlla, we were able to launch a website, ddx3x.org, with a patient registry, form the DDX3X Foundation, and find doctors dedicated to helping to find a cure for this disease. We could not have done any of this without her help and support.

When new families are diagnosed, they all seem to find Karlla first. She connects them to our Facebook page and website, and introduces them to us. And now we estimate that we are up to 100 girls with this disease, though we expect to have many more. Doctors believe the DDX3X mutation will eventually account for up to 3 percent of unexplained intellectual disability in girls.

I feel so fortunate to have found the amazing Karlla Brigatti. Her dedication, support, and compassion are unparalleled in an age when medicine has become so corporate. She is always willing to take a phone call, answer an email, or make a connection — literally even from a Brazilian rainforest.

In my 10-year search for answers, I have never come across another medical professional who is such a generous advocate. We need more people like Karlla Brigatti in this world. She is an unbelievable gift that fell into my family's life, and she has already fundamentally changed the course of many girls' lives thanks to her never-ending generosity.

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