Code Talkers

***Genome* readers honor genetic counselors who interpret complexity with compassion.**

Made possible by

INVITAE

MARGARET

Margaret Au
Open Line of Communication

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On July 28, 2014, just skipping from one foot to the other, my daughter, Amy, fractured her femur. That traumatic moment eventually led our family to Cedars-Sinai and a meeting with genetic counselor Margaret Au. With a 24-year history of rare diagnoses already in her medical chart, Amy's genes had been tested three separate times. Each test had been done in an effort to understand her many atypical health issues, including why her bones were as dense as marble in some places and as thin as cardboard in others. To be honest, our family had little hope that further genetic screening would lead to any new or helpful information. But the traumatic fracture gave us a new sense of urgency to find a unifying diagnosis. The emerging science of whole exome sequencing (WES) offered a new avenue we had not yet explored.

We had come to Margaret and to Cedars-Sinai after Amy's genetic report had shown three mutations not seen in combination before. Even before our first visit, Margaret was our lifeline. Her job could not have been easy. Amy's many issues affected not only her bones but also her brain, eyes, and kidneys, and we had pages and pages of reports from specialists. In fact, the first thing that struck me when talking to Margaret was the sense of relief that someone was really listening as we talked about all of Amy's issues — considering our daughter as a whole person rather than as a list of separate symptoms. Margaret's open line of communication meant that we could call her whenever we had a question or thought of a possible new bit of information that might help. In turn, she helped us understand exactly what genetic testing could and could not do, and what it might mean for not only our affected daughter but also for her sister.

When we had our genetics appointment, Margaret immediately hit it off with Amy, who is cognitively much younger than her years. Margaret seamlessly explained the basics of genetics in words that Amy could understand, and encouraged her to ask questions and contribute her own thoughts to the dialogue.

In a scary new world, Margaret was a constant source of calm guidance. We never felt that any question was too ridiculous, any observation unimportant. For the first time in a long time, we had a partner who was doggedly helping us get to the bottom of this puzzle that is my daughter.

When we had our genetics appointment, Margaret immediately hit it off with Amy, who is cognitively much younger than her years. Margaret seamlessly explained the basics of genetics in words that Amy could understand, and encouraged her to ask questions and contribute her own thoughts to the dialogue. She brought out Amy's goofy personality and made her comfortable throughout an exam that included such strange procedures as measuring ears, examining fingernails, even clipping a sample of hair.

Armed with results from WES and other laboratory tests, Margaret immediately began to consult with the National Institutes of Health (NIH). Due in large part to Margaret's paving the way, Amy was accepted into a study of people with similar genetic profiles, and within a few short months, our family was headed to Washington, D.C. While there, Amy was officially diagnosed with two ultra-rare, life-threatening disorders. Though the diagnoses were daunting, Margaret's care, empathy, and her ability to be the liaison between the doctors and us had prepared us for this confirmation. Likewise, the relationship Margaret made behind the scenes with Amy's sister helped allay fears about her own genetic health by giving her information that will help her to make informed reproductive decisions.

Our journey to diagnosis was long and at times arduous. I am glad that we had the care and compassion of a genetic counselor like Margaret to see us through to the end of the searching and the beginning of treatment. If I had to describe in one word what Margaret gave to our family, it would be empowerment. With her clear explanations and her affirmation that our concerns deserved to be heard, we are more confident caregivers. Though not medical professionals, we know now that our roles as parents, sibling, and patient are equally important. We believe that we can ask informed questions and contribute meaningfully to Amy's medical care. Yes, our genetic counselor has helped our whole family to feel empowered. Thank you, Margaret!

L TO R: JANICE, AMY, MARGARET