Code Talkers

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

Susan Schelley
Finally, a Diagnosis

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My name is Jesse Marimat, and I have a rare genetic blood disorder. At age 24, I was referred by my hematologist to a genetic counselor to help isolate mutations and possibly receive a definitive diagnosis for my disease. I was partnered with senior genetic counselor Susan Schelley to take my case.

She was fascinated by my experience with misdiagnosis and by my comprehensive understanding of genetics. Our counselor-patient dynamic was evidently not the norm. Two slides of my bone marrow were sent to two different research labs.

Over a year, from sample to results, Susan and I kept in constant communication. We were eager to share any new information in the relevant field of genetics.

Susan's compassion and expertise came to light when she received the results from my bone marrow slides. The gene isolated was SEC23B, which was screened for a mutation linked to congenital dyserythropoietic anemia (CDA), a condition I was never confirmed to have, although it was alluded to as I was growing up.

She walked me through the results slowly and with precision to ensure I would understand the implications of the findings, as well as what it meant for future research in the field of genetics and hematology. We found not only that I have the mutation for CDA but that it's most likely the type 2 variant. In addition, I have two novel mutations, on exons 13 and 19, which have not yet been linked to CDA.

Her approach to explaining my diagnosis was professional yet compassionate. She's had to deliver life-altering news to hundreds of patients over the years. Her actions on that day affect my life now, and I reflect back on that moment often.

She was with me on my D-Day — Diagnosis Day. It's the day when a doctor tells you news that confirms your worst fears, or relieves your stress and worry. For me, Susan gave me answers to questions I had been asking myself for two decades. Through her efforts, numerous emails, and hours of discussion, I could finally say with confidence that I had a confirmed diagnosis. That meant the world to me. She helped make sense of the chaos that was my condition, comforted me in my relief at having an answer, and was optimistic about where we go from here.

Given my unique diagnosis of CDA type 2, we are now exploring options for further studies and research related to this particular genetic mutation.

Now 29, I am thankful for Susan's role in helping me define my condition, giving me a purpose, and pursuing more research and development in gene sequencing. Above all, I am thankful for Susan's help in giving me hope for the future.