



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

Made possible by





Gretchen MacCarrick Beacon of Hope

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GRETCHEN

A year ago, I was sitting in the elementary school parking lot with a heavy heart, feeling completely alone. My 7-year-old daughter couldn't walk more than a few steps without becoming completely exhausted. She explained the fatigue as if "my legs were going to fall off." I watched her trudge up the school sidewalk dragging her shoes on the pavement.

I had spent the previous day on the phone, bouncing between the office of the pediatrician and the cardiologist, trying to decide what to do about a heart medication she was on. The side effects were overwhelming, and I was scared to think of how my daughter would be affected.

Out of complete desperation and as a last resort, I reached out across the country to Gretchen. She was someone I trusted and knew could help me find answers. Shortly after the call, our medical team at home had the information that was needed to modify my daughter's dosage and relieve the debilitating side effects.

My daughter has been diagnosed with the rare connective tissue disorder called Loeys-Dietz syndrome (LDS). After receiving the official diagnosis in 2012, I quickly came into contact with Gretchen MacCarrick. I live on the West Coast; she lives on the East Coast. I studied psychology; she studied biology. We are separated by three time zones and more than 3,000 miles.

Gretchen is a genetic counselor at Johns Hopkins Hospital. She works closely with Dr. Harry Dietz, the doctor who is trying to save my daughter's life. Over the past four years, Gretchen has been there to help me make sense of the medical jargon and to set priorities during some of our most difficult times.

She has received many emails with questions like: "What activities can my daughter participate in?" "How often do we need new MRI imaging?" "What emergency procedures should I define for the elementary school my daughter attends?" "Any chance you could help me set up a bunch of specialty appointments there in Baltimore?"

I have quit counting the times that I have hung up the phone after speaking to a medical office and felt completely alone in my daughter's medical care. Through all the researching, phone calls, and trying to get doctors to understand my daughter's syndrome, Gretchen has been on my team. She is someone who believes in me. The belief of another person has been healing and reassuring. Her simple reassurance has been a beacon in a storm of doubt and the unknown.

Most recently, our family moved to a new state even farther away from Johns Hopkins. We started the process of meeting new doctors in a new town. Each introduction to a new doctor included introducing LDS to the doctor.

My daughter's yearly MRI was scheduled shortly after an initial visit. I was encouraged that her care would continue uninterrupted in a new place. Encouragement quickly faded to frustration when I noticed that the MRI had been limited to only her heart and not her entire upper body, including her head and pelvis, as was recommended by the LDS team.

I mentioned this to the doctor, but my request seemed to fall on deaf ears. This syndrome was unfamiliar to the doctor and his staff. This unfamiliarity dissuaded the office from pushing for a complete scan. Not knowing what else to do, I emailed Gretchen for help. Her response came in record time and was simple yet included all the information I needed for my daughter to receive a complete scan.

Although we are miles away, Gretchen has been a consistent source of peace and goodness. She sends emails that reassure and deliver hope. More than once, a simple note from her has given me the strength to continue working with doctors to find the best care for my daughter. She not only cares for the patient, she cares about the patient. She is upbeat and optimistic in helping us to find our own source of optimism. Gretchen has dedicated her life to improving the lives of others.

A mother's love runs deep for her child. I am forever grateful for the countless hours Gretchen has spent on my daughter's behalf. ☺

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