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All in the Family

A health crisis can spawn the ultimate application of biotechnology—attempting to save a loved one's life

BY RENEE MORAD

Doctors diagnosed Karen Aiach's daughter Ornella, at just six months old, with a rare condition called Sanfilippo syndrome, in which a missing enzyme causes toxic substances to build up in the body. Aiach began educating herself on everything from the disease's symptoms and outlook to its leading physicians and researchers. "With information so readily available these days, the wall between patient and their caregivers and doctor is diminishing," Aiach says. "I learned which authors were publishing research studies and felt empowered to pick up the phone and contact them to learn as much as I could, as quickly as I could." Aiach's research motivated her to leave her job running a consulting company and start Lysogene, a biotechnology firm in France that is currently developing gene therapy to treat Ornella's syndrome and other neurodegenerative diseases.

When faced with a loved one's difficult diagnosis, most people describe feelings of being jolted into a race against time. I recently caught up with a group of entrepreneurs who have all lived through this experience and gone on to create remarkable biotech companies to improve outcomes for their family members and others. Confronted with a life-or-death situation, these individuals didn't just hope or yearn for good news, they created their own—with the help of a carefully enlisted team of strategic partners. Their biotechnology-related companies and organizations, though inspired by the plight of a loved one, went on to enrich the lives of dozens, hundreds, and in some cases, thousands of individuals facing similar obstacles.

ADJUSTING A GENE

Patients with Sanfilippo syndrome develop mentally and physically up to age four, then lose what they have learned, while suffering from sleep disorders and hyperactivity. The median life expectancy is 15 to 18 years of age. Aiach's initial research revealed that few treatments were in progress for this syndrome and that gene therapy would be the most promising course to take. "I set out to find a therapeutic approach to the disease to make life more manageable for my daughter and others struggling with the syndrome," Aiach says.

Lacking a medical background, Aiach hired a key neurobiologist and other staff members to help lead the way. Just a few years after the company's launch in 2009, Lysogene conducted Phase I/Phase II clinical trials of an experimental treatment. The genetic therapy has gone on to make a positive impact on Ornella's day-to-day wellbeing. "She's quieter and calmer now, not enduring as severe bouts of hyperactivity, and is leading an overall happier life," Aiach says. Although it's still a work in progress, Aiach is confident Lysogene is moving in the right direction.

A POWERFUL ONLINE COMMUNITY

In 1999, Jamie Heywood received devastating news that his brother Stephen had been diagnosed with amyotrophic lateral sclerosis (ALS). The crisis, Heywood says, immediately brought his already tight-knit family together to "an amazing level of closeness and collaboration." His family teamed up in numerous ways to help Stephen face his condition and to give him a fighting chance by gaining access to the best doctors. They also set out on a mission to help him experience the highest possible quality of life.

"We immediately started building things, because that's what my family does," Heywood says. That meant extensive remodeling projects—which was a passion of Stephen's—to keep him motivated and thriving. It meant founding the ALS Therapy Development Institute, the world's first nonprofit biotechnology company, dedicated to ALS treatment. It also meant starting PatientsLikeMe, an online network where individuals share information about their illnesses, symptoms, treatments, side effects, outlook and more.

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Launched in 2006 as a virtual community for people with ALS, PatientsLikeMe later expanded to include patients with other conditions, such as multiple sclerosis, Parkinson's disease, chronic fatigue syndrome, epilepsy and organ transplantation. Today its successes range from singular victories—like connecting a patient with epilepsy with an epileptologist who was able to provide life-changing treatment—to large-scale projects, such as gathering and analyzing patient-reported data to aid in drawing big-picture conclusions about drug safety. "In this community, patients are empowered to share everything that's worked, everything that hasn't worked, their best advice and their biggest fears—and it all comes together to provide a very powerful platform for patients," Heywood says. "It's about gathering experiences and data that can help accelerate research and provide better treatments." There's also that sense of community that reminds patients that they are not alone in their experiences. Heywood's brother's life was unfortunately taken much too soon by ALS, but the PatientsLikeMe online community continues to shine as a beacon of hope and positivity for hundreds of thousands of others.

RESEARCHING A VERY RARE DISEASE

Matt Wilsey and his wife Kristen settled for nothing short of comprehensive when tackling their daughter Grace's illness. Wilsey explains that he and his wife immediately knew something was wrong at his daughter's birth, due in large part to their baby's floppy and lethargic nature. So the family traveled around the country to hear the opinions of top doctors, and had whole genome sequencing conducted at two different centers to see if they yielded the same answers. They did, and after some trial and error, Grace was diagnosed as the second American and the sixth patient worldwide to have a rare genetic disorder known as NGLY1 deficiency, which causes severe brain, liver and muscle problems.

To advance treatment for their daughter's disease, the Wilseys started the Grace Science Foundation, and soon plan to launch a for-profit biotech company with a global team of 75 researchers. "Instead of sitting back and waiting while a large pharmaceutical company keeps this disorder as number 1,000 on a list of potential diseases to address, we decided that if we have the bandwidth, resources and scientific chops, we can get to work and perhaps explore partnering with a larger company in the future," Wilsey says.

In this way, the Wilseys hope to bring new options to Grace and to patients like her around the globe.