To Save the Children

BY JOHN CROWLEY, '92 J.D.

"Go ahead and enjoy the time you have with your children," we were told. In just one hour, our world was changed by circumstances that we never could have imagined, and that, we were told, we could not change.

I remember the first time I saw the Notre Dame football team play on television. It was January 1, 1975. My dad, a New Jersey police officer, had regaled me with stories of the legends of Irish football, and on that chilly, January night, we were ready to watch our first game together as father and son. It was a right of passage, of sorts, in our family. I was 8 years old. I still remember being mesmerized by the glow of the stadium lights off the gold helmets. ND beat Alabama in that Orange Bowl, 13-11. After the game, I asked my Dad for a helmet just like the one the ND players donned. The next day, we went to Sears, bought a generic white football helmet and spray-painted it gold. Twelve days later my Dad, my hero, died in the line of duty. He was 35. This experience would forever change me. I would learn to cherish each and every day with those I love. I would resolve to deal with each and every challenge in a way that would make my Dad proud, and I would one day get a degree from the University of Notre Dame.

In 1990, my high school sweetheart, Aileen, and I married. We moved that summer into our first apartment, in South Bend, Ind. I was a second-year law student at Notre Dame. The three years studying law at ND taught me what it means to be a citizen and, most importantly, about choosing right over wrong, especially when "right" seemed so difficult. This foundation would serve us well in the years to come.

After practicing law in Indianapolis for four years and later earning an MBA at Harvard, Aileen and I moved to Walnut Creek, Calif. We were on top of the world. It was 1997. I had a great job with a consulting firm in San Francisco. Most importantly, we had two beautiful children. John was two, and Megan was one. By March of the following year, we were expecting our third child. Three kids aged three and under—the perfect Irish Catholic family was in the making! In the months before our third child Patrick was born, however, we began to notice that Megan wasn't as strong as a one-year-old child should be. She wasn't pulling up in her crib, and she wasn't taking her first steps. The doctor recommended that

John and Aileen and their children (from left to right), Megan, John, and Patrick, enjoy time together as a family despite the demands of pursuing a treatment for the youngest children's rare disease.

she have a biopsy so they could examine her muscle pathology and determine exactly what any problem might be.

Patrick came to us on March 13, 1998. He was a great big, healthy newborn, an Irish lineman-to-be, no doubt. Seven days later, the physician in California had the biopsy results on Megan. In a steady but nervous voice, he explained that the results showed that Megan's muscles were rapidly accumulating stored sugar in the form of a substance called glycogen. This glycogen accumulation was leading to the rapid degeneration of all of her skeletal muscles and her heart muscle. She had a disease called Pompe Disease. Aileen and I had never heard of this strange disease. We were told that there was no treatment and that babies diagnosed with Pompe rarely lived past age two. She was 15 months old when we received the diagnosis. The physician went on to tell us that Patrick, only seven days old, had a 25 percent chance of having the disease, as well.

It was a long ride home from the hospital. Aileen and I were both very healthy and had no family history of any disease. Pompe is a rare, recessive disease. Parents are carriers and have no symptoms and usually no family history for the disorder.

The doctors that day told us that there was nothing we could do, and that we should accept this most difficult of diagnoses. "Go ahead and enjoy the time you have with your children," we were told. In just one hour, our world was changed by circumstances that we never could have imagined, and that, we were told, we could not change. Throughout that day, Aileen, our close family, and I went through a range of emotions. We experienced shock, denial, anger, sadness—and fear. We feared for Megan and possibly for Patrick, too. (Our oldest son John, thankfully, was already a thriving and strong three-year-old, without Pompe). We knew little of the medical world. The Internet 10 years ago was just taking hold, and I spent that night on the computer to learn about this strange disease and to see

if there was anyone, anywhere, who might be able to help our baby girl. I put into Yahoo the words "Pompe Disease," and with that, stepped into a whole new world of diseases, biotechnology, and medical science.

Pompe disease, I learned that first night in front of the computer, was first characterized in 1932 by Dutch pathologist Dr. J.C. Pompe. He was the first to make the connection between the glycogen coming from a deceased baby's muscle cells and the symptoms of the disease—the enlarged heart and weakening of the muscles that eventually led to death. I learned, too, that researchers in the Netherlands and at Duke University were working on competing projects to develop an enzyme-replacement therapy in their labs that could be used in humans with Pompe. The thought was that with this lab-made enzyme, we could replace the ineffective enzyme that patients with Pompe naturally made. We could, in essence, use this biotechnology to treat the disease. After reading and learning for hours, I woke Aileen to tell her about all that I had discovered. Still groggy from her deep sleep, and just seven days after a third C-section, she asked me: "What does this all mean?" I responded, "It means hope, Aileen. It means maybe there's hope."

Two months after Megan's diagnosis, we got the results of testing on Patrick. To our great surprise and sadness, Patrick was also diagnosed with Pompe. By the fall of 1998, within just six months of her diagnosis, our little Megan rapidly deteriorated. She lost so much strength that, when she got a simple cold in September, she developed pneumonia and lost the ability to breath on her own. She ended up in the pediatric intensive care unit (PICU) of a local hospital here in New Jersey. For hours, the doctors struggled to stabilize our 20-month baby girl. We were told to expect the worst, and we just prayed that morning, that if that was going to be the outcome for Megan, that it come quickly. "Please, God, don't let her suffer any more."

After hours of waiting, I recall vividly when the head of the intensive care unit came into the waiting room. Aileen and I embraced, waiting to hear the words that we dreaded. Instead, he told us that Megan was breathing on a ventilator and stable. The doctor was amazed. He told us we had some little fighter—that she refused to give up—and that we could briefly see her. We entered the PICU and we found Megan lying still and awake on the bed, medical tubes coursing through seemingly every vein in her body. Her eyes were moving around the room, inspecting all of the action, and finally locked on to her mother's eyes, with both bursting into tears. It was in that moment that we knew that she didn't want to quit—and neither could we. I stroked her brown hair and told her, "Okay, Princess, if you want to fight, we'll fight, too."

Over the next few months and years, our lives would continue to change and to be defined by a quest to help our children. By early 2000, we had grown frustrated with the pace of research in Pompe. A treatment always seemed right around the corner, but never came. Finally, in March of 2000, I left my job at Bristol-Myers Squibb to take over as the CEO of a four-person biotechnology start-up company based in Oklahoma City that was doing research into a new medicine for Pompe. The company was called Novazyme, and over the next 18 months, we grew Novazyme into a 120-person company. Admittedly, I knew little about the world of biotech and venture capital; to say that we learned "on the job" is even being generous. Still, we had a vision and a mission, and failure was not an option. Novazyme was eventually merged into one of the world's largest biotechs, Genzyme Corporation. At Genzyme, we continued the work to develop a medicine to save, extend, and enhance the lives of people with Pompe, and I struggled to get my own two kids access to this experimental treatment. Even this proved to be a momentous struggle against nature, time, and even accusations of conflict of interest. It strained our lives, our marriage, and our friendships. Finally, nearly five years after their diagnoses, Megan and Patrick received the

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enzyme-replacement therapy that they so desperately needed. It was January 9, 2003. That day would have been my Dad's 63rd birthday. Somewhere, an Irish cop was smiling in heaven.

The enzyme therapy has saved the kids' lives. Their hearts, which had swollen to nearly twice normal size, within months returned to normal. It was our miracle—and they are our little miracles. Today, over 500 kids worldwide with Pompe take this enzyme, now an approved drug known as Myozyme.™ Even with this drug, Megan and Patrick are still profoundly physically handicapped from the ravages of the disease on their skeletal muscles over the years. We continue the search for new and better ways to treat them and to make them stronger. The advances of the past decade are the results of the efforts and passion of hundreds of scientists, businesspersons, caregivers, patients, and their families. Megan and Patrick's courage and inner strength have taught us more about love and life than we could ever have taught them over these years. And so we grab onto each precious moment with them, cherish it, celebrate it, laugh at it, cry in it, and hope for another, even as we continue the journey into the unknown and unknowable that we call life.

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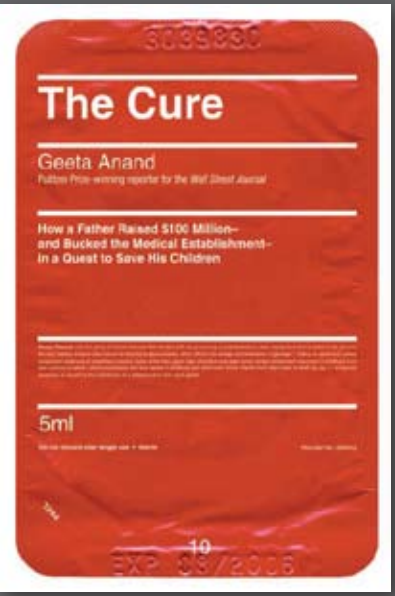
John Crowley is president and CEO of Amicus Therapeutics, Inc. He is also a commissioned officer in the US Navy Reserve and has just recently completed a six-month active duty assignment.

While a student at the Law School, he was a member of the national moot court team and was named best advocate in the showcase argument before Supreme Court Justice Kennedy and two other judges. He was selected to deliver his class' commencement address.

John and his wife, Aileen, and their children, John Jr., Megan, and Patrick, live in New Jersey.



The Crowley family story is told in a book published last year called ***The Cure: How a Father Raised $100 Million—And Bucked the Medical Establishment—in a Quest to Save His Children.*** It is authored by Geeta Anand, a Pulitzer Prize-winning reporter for ***The Wall Street Journal.*** The Crowley family story is also being made into a major motion picture by Sony films, produced by Michael Shamburg and Stacey Sher (producers of ***World Trade Center*** and ***Erin Brockovich.***) You can learn more at: **www.thecurebook.com** or **www.crowleyfamily5.com**.