CRITICAL PATH INSTITUTE

Given No Answers After a Life-Changing Diagnosis, Pat Furlong Worked to Create Her Own

By Alexander Diegel

It was 40 years ago that Pat Furlong faced a heart-wrenching diagnosis for any parent; her sons Christopher and Patrick were diagnosed with a grave and fatal illness, Duchenne muscular dystrophy (DMD). DMD, a rare and severe form of inherited muscular dystrophy, was first diagnosed in Christopher after he suffered an Achilles injury from trying to ride a bike. The doctors previously told her that concerns over his slow development were merely parental paranoia, and that he would catch up. They were wrong.

The recommended treatment at the time was therapeutic nihilism. In other words, go home, cry, and enjoy your remaining time with your children, both of whom died in their teens. But Furlong, who has a master's degree in nursing, found this "treatment plan" to be unacceptable. So, she started a movement to find a better plan that, even if it couldn't save her own sons, would save, extend, and improve lives for future generations of boys diagnosed with DMD. While females can be carriers and pass down the disease, it is inherited in an X-linked recessive manner, meaning males are overwhelmingly more affected than females.

After quickly learning that researchers and physicians didn't want to talk to her, because parents were considered desperate and uneducated, she falsified her identity in order to visit some of the top laboratories across the country and talk with researchers.

"I promised myself I wouldn't cry, and made appointments at major labs with Harvard, Yale, Penn, UCLA, Stanford, and I made up that I was a postdoc looking for a job," she explained. "I wanted to understand what do we know and what don't we know [about Duchenne]? And I wanted to understand who the players were. So, I travelled around and began to understand, 'Okay, there's no vehicle at the moment to even develop a therapy and get it approved."

Little did Furlong know it at the time, but her incognito interviews were just the beginning of a lifetime of advocacy work she would dedicate to all people affected by Duchenne. Ten years later, Furlong founded Parent Project Muscular Dystrophy (PPMD), where she still serves as President and CEO and has been a catalyst for most major changes and developments in therapies for DMD.

For the last 30 years, PPMD has invested in every single therapeutic development for Duchenne. The nonprofit takes a cutting-edge approach to accelerate the development of treatments that will someday end DMD for every person and family impacted by the disease. PPMD has invested more than $55 million into Duchenne research, contributions that have led to advancements that inspired an additional $600 million investment in research from the federal government.

20,000

Children Are Diagnosed with Duchenne Each Year

15,000

People Are Living with Duchenne in the U.S.

300,000

People Are Living with Duchenne Worldwide

In trying to advance treatments and develop better, safer medicine for these young boys diagnosed with DMD, Furlong grew frustrated with how isolated the data was. There would be data from one particular registry, then another group would have another registry, and that was separate from other important datasets. It was around 2016, when she was collaborating with FDA's leadership in neuroscience, that Furlong was first introduced to C-Path and decided to work together to establish C-Path's Duchenne Regulatory Consortium (D-RSC).

D-RSC breaks down the silos Furlong experienced to advance treatments for DMD through support of collaborative research and shared data access. The PPMD founder has since stayed involved with D-RSC as a co-director, where she has co-authored papers with the C-Path team, attends conferences, supports D-RSC's portfolio activities and is a regular speaker at C-Path and D-RSC events.

"We have been frustrated, knowing what integrated data could do for us in terms of research and drug development in our community. Plus, the process takes so long, and time matters. Time is muscle. Time is life for us," Furlong said. "What C-Path does is approach all the key players for their data, and gets that data released and integrated for the community to process."

"Pat's unwavering dedication and heartfelt advocacy have cast a transformative light in the DMD community," said C-Path's D-RSC Executive Director Ramona Belfiore Oshan. "Her leadership and vision have been instrumental for D-RSC's mission of aligning groundbreaking research and enhanced drug development tools with the patient community perspective and needs."

While it's been a long struggle and there's still a way to go before treatments advance to make Duchenne a manageable disease with a favorable long-term outcome, there has been significant progress. Boys are now growing to men, reaching adulthood, having successful careers and inspiring hope for parents whose children have just been diagnosed with DMD.

With more collaboration among industry, academia, regulators, patient groups and the DMD community, we expect more data — and more reason to hope for treatment advancements in the years to come.

"I think all of these data that C-Path has and will have in the future is going to lead us to a future for these young men, where they walk for a very long time, hopefully forever, and their hearts are stable and where insurers are covering the therapies that are recommended," Furlong explained. "We believe that Duchenne, and those living with it, have a future."

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