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INSIGHTS

Family reflections: Duchenne Muscular Dystrophy

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The past 2 years in the midst of the coronavirus have been a trying time for all of us, but more so for families with a member suffering from an existing, life-limiting health issue.

For Duchenne Muscular Dystrophy parents like myself, this time has been particularly agonising. When facing a disease where time is a luxury, it is of the utmost importance that science moves as fast as safely possible in the hopes that a cure might finally be found for our children.

However, as the pandemic spread, we were forced to reckon with the fact that much promising research, potential breakthroughs and a possible 'Eureka' moment were all pushed to the back of the table while scientists rushed to create a vaccine and treatments for the pandemic.

Now that Duchenne research is slowly coming back on track, us Duchenne parents have finally been given back the one thing that keeps us moving forward: hope.

When my son, Hermes, was diagnosed in 2012 at the age of 13 months, my first question after being given the diagnosis was "What's the therapy?" only to be told that there was none. It seemed unbelievable. How could we be in the twenty-first century without treatments for certain diseases? Over the following years, I was to realise that this was the reality for most rare disease patients.

Duchenne is a complex and intricate disease. If its outcomes weren't so devastating, you might even call it fascinating. Tiny fragments on a specific gene in the body are missing, duplicated or otherwise malfunctioning, and this causes a catastrophic failure in how our childrens' muscles work.

Most of us never heard of the term before our children were diagnosed. It is not a glamorous disease to be working on as a scientist, if such a term can be used. It is a quieter condition, one where the perhaps the boldest and most ambitious researchers dare to push for new frontiers.

Duchenne is a rare disease with a small patient body. The trials that can be run are limited for this very reason. The successes are few and the failures are many. Currently, the only protocol is to administer steroids, which comes with its own litany of side effects.

My hope is for there to eventually be a genetic therapy, but I am well aware of how much of a gargantuan task this is, taking on the tiny broken fragments of our children's genes and trying to work out how to give them a better tomorrow that us parents appreciate so much. It gives us a reason to look to the future and hope that it might be better, despite knowing that physically for our children, each of our yesterdays is better than all of our tomorrows.

The quiet, persistent efforts of research towards giving our children better lives is invaluable in a world where time seems to be speeding forward when we as Duchenne families don't have a moment to take for granted. It means that the light at the end of the tunnel that was hijacked by the pandemic is back in place now and that if not for our childrens' generation then for the next, Duchenne might not be a death sentence.

Thanks to the tireless work of researchers, we are already discussing how Duchenne Muscular Dystrophy can be managed as a chronic condition when a few short years ago, adult patients were a minority so small, no consideration had been given to how to manage their lives.

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Scientifically sound research also adds an extra layer of protection against another distressing phenomenon in the sphere of rare disease. Unfortunately, desperate parents will look for any solution, and this desperation is often exploited by charlatans peddling their snake oils. When there is little hope, desperation pushes you towards unsound choices and there is no shortage of salespeople waiting in the wings to pounce on such people. This is no less true with Duchenne Muscular Dystrophy, where sadly, year after year we are obliged to flag up a new catalogue of untried and untested therapies being pushed on desperate parents and carrying eye-wateringly high-price tags.

Research gives us more to look towards while we wait for a cure for this condition and comforts us that the future might not be so bleak for our children. Respiratory and cardiac care, psychological care, digestive issues and more are all peripheral issues that can be worked on to improve lives and outcomes and reassure parents that their children can still have meaningful and physically comfortable lives.

In the Greek myth, Pandora was created by the gods and given a box before being sent among humanity, where she opened the box, releasing a litany of sorrows, sickness and evil on humans. As she rushed to close the box again, she trapped one thing at the bottom—hope.

With the fear, illness and sadness that Duchenne families face, research into improving outcomes for our children is akin to the step by step resealing of that box to hold within it the hope that keeps all of us going.

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

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