Pediatric RESEARCH

INSIGHTS

Family Reflections: MPS IVA- Morquio Syndrome

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Scott is 17 years old, living with Mucopolysaccharidosis IVA (MPS IVA) also known as Morquio Syndrome. He recently graduated from high school and will be attending the University of Washington in the fall to study biomedical engineering.

We would like to share our appreciation for everyone involved in his care, especially researchers who work tirelessly to ensure the best possible outcome for those with Morquio Syndrome.

MARY (SCOTT'S MOM)

After 3 years of searching for answers to Scott's health issues, I clearly remember the doctor's words, "It's been confirmed. Your son has Mucopolysaccharidosis type IVA – Morquio Syndrome."

Without hardly taking a breath, he continued, "But there is a treatment being tested and the clinical trial will be recruiting patients shortly."

I did not know it at the time but discussing an actual treatment while presenting a rare disease diagnosis is very uncommon.

While we immediately understood the importance of having a treatment for a progressive disease, it was not until later that we discovered we were part of a lucky few.

We flew once a week from Seattle, WA to Oakland, CA for a year and a half to be part of a clinical trial for MPS IVA. Scott missed over 80 days of school and spent at least 1 day a week in the hospital, sometimes a week at a time for medical testing. The treatment was a weekly IV infusion that lasts more than 5 h from start to finish and it is for life. We eagerly accepted this inconvenience and stress for the chance of a better life for Scott.

Before treatment, Scott was having a lot of pain and headaches. I found myself picking him up from school more and more instead of letting him walk home. We started giving him more NSAIDs to cope with pain from activity such as playing soccer, ice skating, or simply going out with friends and walking around.

After going on treatment, changes started to happen. They started so slowly that we did not really notice anything until after he was on treatment for 6 months. Then all of a sudden it occurred to us that we were giving him less and less medication, he was complaining of less headaches and seemed to have more energy. After only hoping for the progression of the disease to slow down or stop, we were thrilled to see actual improvements. To this day, it is quite evident to us that we owe his current physical condition to this medicine.

At the end of the trial, we were invited to Washington D.C. to testify at the FDA Advisory Panel. We had the pleasure of meeting some of the members of the pharmaceutical team and those responsible for taking the enormous risk of bringing this treatment to market. It was a pleasure to meet the people that were trying to change the course of this disease and thus changing Scott's life and also the lives of our family members.

The researchers that studied Morquio Syndrome and developed the treatment are, in our opinion, remarkable. They could have chosen to work on a much higher profile disease, such as heart disease or cancer. New treatments for these diseases are publicized widely in the media and published in multiple scientific journals. The recognition is far reaching. Those who chose a niche area celebrate the accomplishments in a more subdued and less public manner.

In addition, ignoring rare diseases has a large impact economically on America; the opportunity cost of lost wages and productivity for patients and their unpaid care givers, the cost to Medicare, Medicaid and Social Security programs, and the significant medical costs of managing the symptoms and complications of diseases with no treatment. Many recognize that a society can be measured by how we treat those who need the help of others.

Much of the research into rare diseases can be translated into research for more common diseases. Most rare disease are caused by a mutation of a specific gene whereas most common diseases involve multiple genes and environmental factors and are therefore difficult to understand. Rare disease research is invaluable in the understanding of all disease processes.

I can assure you that when Scott was diagnosed, we did not care how rare or common the disease was. It really did not matter to us if there were 500 or 5 million Americans with this disease. We wanted someone to find a cure for it.

No disease is too rare. No individual should be forgotten.

For all the researchers, scientists, and physicians who choose to dedicate their careers to rare diseases, we thank you from the bottom of our hearts. Thank you for not shying away from the many challenges of researching a rare disease; from securing financial investment, working with limited natural history data, difficulties in recruiting for and designing clinical trials, and for the potentially enormous monetary and professional risk. We recognize that it is also particularly difficult in this current climate where pharmaceutical companies are continually being called out for high drug prices and high profits.

As I watched my son walk down the aisle at his high school graduation ceremony a few days ago, I silently thanked everyone involved in his care. Blessing the researchers who forged ahead with developing a treatment for a patient base of only 2000 individuals worldwide. I wondered if, without his treatment, would he have been able to walk down that aisle, unassisted, standing proud with his cap and gown, and looking forward to a bright future ahead.

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SCOTT

I am extremely happy with my health and my life right now and it is all thanks to the amazing researchers who have provided me with a treatment. I have Mucopolysaccharidosis IVA (MPS IVA) which is an extremely rare disease with only 500 patients in the United States. I am one of the fortunate 5% of rare disease patients who have a treatment for their disease.

When given a choice between helping a few individuals or helping a large population, many people believe it's admirable to do what's best for the greater good. It means the world to me that researchers have spent countless hours working on a treatment for a disease as rare as my own. They could be working to treat a larger market of patients who have a more common disease, but they choose to remember that my small community matters.

10% of the population, or approximately 30 million Americans, have a rare disease. Therefore, rare diseases are very common in the aggregate. 30 million Americans should not be left on their own, without any research into their disease taking place and therefore left with no hope for a treatment or cure.

I would like to thank these researchers for making my life incredibly better than it would have been without a treatment. Today, I am able to walk around with my friends, go biking and swimming, and even play some pickup sports. Having this treatment allows me to perform to the best of my ability at school without being in constant pain. I have no doubt in my mind that this treatment has changed my life for the better.

Having a treatment has not only affected me but has also changed the lives of my entire family. We do many activities as a family that otherwise would not be possible. My parents do not have to worry that my future will be filled with the pain and struggle that would be certain without a treatment. I can now live a near normal life and dream of a peaceful future.

I would like to pay it forward as I start college this fall. I am interested in studying biomedical engineering so I can eventually make hospital visits easier for children with medical issues and all those with rare diseases.

BEN (SCOTT'S YOUNGER BROTHER)

It is quite amazing how much things have changed over the past few years because of Scott's treatment. I remember Scott coming home many days with a headache and not being able to do anything at all, including on one of his birthdays. Our family was often held back from doing fun activities, such as going on long bike rides, due to my brother's condition. It was also hard when my mom and brother were gone to California for 2 days every week for 1 ½ years.

When Scott started treatment, I honestly did not notice anything at first. It was not until I looked back that I realized how much better things were. Scott was able to do more things with our family and his friends. Now, if our family wants to go on a bike ride, there is nothing stopping us.

For the last few years, Scott has been able to have his treatment at home. I hardly notice when it is infusion day. It is now simply part of our life.

All of the effort from my brother, my mom and dad, the researchers, and doctors has truly paid off and helped our family. I would like to thank all of the hard-working and dedicated people for everything that they have done for us.