Patient and Caregiver Perspectives from the 2023 NORD Breakthrough Summit

We asked those impacted by rare disease to share their experiences and takeaways from the summit.

By Eric Monticello

It's often said that rare diseases aren't that rare. And the National Organization for Rare Diseases (NORD) Breakthrough Summit gives patients a chance to see that for themselves.
This year, 900 rare disease community members came together in person to network, share stories and gather information about thousands of rare conditions. CSL, whose medicines treat rare
diseases, attended the summit and shared information about bleeding disorders and policy changes that could benefit the patient community.

What did attendees representing various rare disease communities take away from the summit? We interviewed them to find out.

JoBeth Souza, President of the XLH Network and mother of a daughter with X-linked hypophosphatemia (XLH).
This was my first opportunity to attend the NORD summit because of COVID and funding within our organization. My first impression of NORD was the size of the conference and the number of individuals in attendance. Their reach is extensive and certainly inclusive. The organization clearly communicates their support for rare disorder organizations through programming, community access to grants and scholarships and resource commitment like the new modules for orphan drug FDA approval.

I am a firm believer that being in the room with other rare disease organizations and representatives is priceless. The information exchange that occurs is incredible, the brainstorming is helpful and the support exchanged is comforting.

An unexpected surprise was seeing the exhibitors, especially the Platelet Disorder Support Association (PDSA). I had a difficult summer being diagnosed with my own rare disease. I was diagnosed with ITP, or primary immune thrombocytopenia, which is a rare hematological autoimmune disorder where the body mounts an immune response towards platelets and destroys them. I started having issues at the beginning of the year and was diagnosed in May 2023. I was shocked to see the PDSA at the conference. I had no idea that ITP is considered a rare disease.

I was so busy trying to stabilize my platelet count over the summer, it was difficult to find time to research ITP. And there seemed to be limited information available on the internet. I truly learned more about ITP in the two days that I was at the NORD conference than I did all summer. For that, I am incredibly thankful to NORD.

Lily Emmanuel, NORD Running for Rare Community Partner, student and rare disease patient advocate living with hypermobile Ehlers-Danlos syndrome (hEDS).
The NORD Breakthrough Summit has benefited me in many different ways. It gave me the opportunity to make connections with people of all different ages, educational backgrounds and connections to rare diseases. Everyone I met treated our differences with curiosity and it created a very safe and comfortable atmosphere. I was able to make new friends, share my story and learn from others. I received so much insight and advice in preparation for entering college next fall, finding a career path and how to advocate effectively.

This was also my first time navigating my medical needs while traveling alone. It was extremely nerve wracking and something I would not have tried without the support of NORD. Even just through the journey to this summit, I learned more about my needs as a rare disease patient and about independence. Overall, I gained a lot of confidence for moving forward into adulthood.

Marla Chapleau, Representative of both Alliance MLC (megalencephalic leukoencephalopathy with subcortical cysts) and the United Leukodystrophy Foundation (ULF), pharmacist and mother of a rare disease patient.
I truly wasn't sure what I would learn and take back home with me to Ontario, Canada, from this conference. I attended alongside my husband, Ron, who is the current president of the ULF. While he was busy maintaining and making connections with many pharmaceutical companies and other interest groups, I too was busy meeting many people and sitting in on some thought-provoking seminars. With a diverse set of topics ranging from the perspective of the caregivers to the U.S. Food and Drug Administration (FDA) perspective on the development of orphan drugs, to talking about the ever-important topic of mental health, people were sure to find something that piqued their interest.

I was happy to sit in on the repurposing of medications seminar as that makes so much sense from an economics point of view. What inspired me the most at this conference was seeing a very different side of the pharmaceutical industry, contrary to how they are frequently portrayed in the media and pop culture. I was impressed with their sincere compassion for those affected by rare diseases and their caregivers. They showed a willingness to spend time, money and expertise in order to help those suffering find a better path forward.

As a pharmacist, I am fully aware that for many pharmaceutical companies, supporting a clinical trial for a rare disease may not fit their end goal. Let's face it, rare diseases will not produce huge profits due to the size of the population affected. I am amazed and grateful to learn that there are some biotech companies that are willing to put themselves out there. As a mother of a 25-year-old suffering from a progressive neurological genetic disease, I wish to thank them.

Another fact that quickly came to mind was that there was such a multitude of people from diverse backgrounds with many people being there because they have been directly impacted by a rare disease (self, family, friend). Though each of our rare and ultra rare diseases are unique and most of us have not heard of many of the other rare diseases, it is truly comforting to realize we truly are not alone.

Allison Robison, Student and rare disease patient advocate living with cryopyrin associated periodic syndrome (CAPS).
This year I was able to attend the 2023 NORD Breakthrough Summit via the NORD Students for Rare program. As an individual who has an ultra-rare genetic condition it is extremely inspiring and hopeful to see all the work going on within the rare disease community.

While this is very positive, sometimes it can be frustrating to see some overrepresentation of programs and opportunities for "rare" conditions and less representation of "ultra-rare" conditions. Despite this, attending the 2023 NORD Breakthrough Summit was extremely helpful in learning about other rare conditions, advocacy efforts, bridging the gap between provider-patient-insurance and the FDA; as well as specialty pharmacies, programs and trials for rare patients and a sense of community through the amazing NORD Students for Rare Program.

Each year, more is being done within and for the entire rare community and that is a beautiful thing. The rare community is one large family that really understands each other – so having the ability to network and connect with other rare community members, whether it is caregivers, rare providers, patients, parents, siblings, or even rare industry pharmaceutical members through the summit is truly invaluable.

Laura Bonnell, President of the Bonnell Foundation: Living with Cystic Fibrosis, and mother of two daughters with cystic fibrosis.
At the NORD summit, I met someone from Iceland who lost a child to disease and is now an advocate. I learned there are only eight people in Iceland with cystic fibrosis. I also met a mom whose daughter has a rare disease and she had to fight for research and drugs, and on and on.

When I heard experts speak at the event, I realized in some cases I knew as much, or even more than some of them. That's not a bad thing. My point is we are all immersed in rare diseases. We all have insight. We all have something to say. We can offer hope and share information.

The roundtables, workshops, networking, speakers and presentations were incredibly important. It got us together, sharing ideas and contacts. On leaving the NORD summit, I felt I learned from everyone I came in contact with and was able to offer some answers and connections to others. I also felt like we are an important community that is looking out for the entire country, whether they have "rare" in their life or not. We're working to make legislation better for all.

At the summit, CSL's Scott Hambaugh participated in a panel discussion on clinical trials for orphan diseases. A team also shared a CSL-sponsored report on the treatment and management of inherited bleeding disorders. The report urges patient involvement as well as regulatory, health technology assessment and payment models that support innovative treatments.

"At CSL, we are committed to supporting patient access through responsible pricing principles and solution-oriented contracting models to help governments manage budget impacts, continuous long term data generation on the value of our therapies, best-in-class patient assistance programs and therapy donations," said Mary Lacey Reuther, CSL's Head of North America Policy & External Affairs.