Julie R. (New Jersey):

What is your connection to rare disease?

My son Ben was born with congenital hyperinsulinism (HI), a condition that causes the overproduction of insulin that causes prolonged and severe hypoglycemia, a serious disease that has no approved treatment for many of its subtypes. Despite the diagnosis, my husband Mark and I were determined to give Ben the best possible life. After meeting people all over the world who also had children with the disease, it reinforced for me the need to join with others to improve the lives of these children wherever they were born. For that reason, in 2005, I joined with other parents of children who were also affected by the condition to found Congenital Hyperinsulinism International (CHI).

Since 2010, I have been the executive director of CHI. CHI has created an active worldwide community of patients, their families and caregivers, expert clinicians and researchers, and professionals in the biotech field – to fulfil CHI's mission to find better treatments, prevent death and brain damage, and support HI families every step of the way.

CHI has developed and launched the HI Global Registry, a global patient powered research project, developed the CHI Collaborative Research Network with a global group of experts and patient leaders, secured funding for many pilot research grants, supported patients with emergency funds, medication donations, targeted genetic testing, and support groups. CHI has organized many global conferences, created disease awareness information in 23 languages, and provided patient experience expertise to six biotechs.

What are you hoping to accomplish by advocating and volunteering with NORD?

I am very interested in a large number of advocacy issues. In my mind, most pressing are advocacy issues that help decrease the time to diagnosis, as many longstanding medical issues that result from having a rare disease can be ameliorated or averted if diagnosis occurs early in the disease's onset. Therefore, newborn screening is very important to me. We want every newborn to have access to screening for all the diseases that are on the national Recommended Uniform Screening Panel. In addition to the screening of the treatable diseases currently included in the panel, I am very interested in legislation that would increase how we screen, and add more diseases to newborn screening, whether this is done through more genetic testing, or adding new ways to screen for rare but treatable diseases.

As an example of the current newborn screening limitations, the condition my son was born with, congenital hyperinsulinism, can cause brain damage or death if it is not diagnosed soon after birth. A drop of blood taken soon after birth and tested for glucose level could being the process of saving a life or preventing brain damage, but since newborn screening relies on a dried blood spot, and there is no way of detecting the condition with a dried blood spot, congenital hyperinsulinism cannot be added to

the tradition newborn screening program. The same may be true for many other diseases. Therefore, expanding how and what we screen for in the newborn period is of great interest to me, as well as legislation to make it happen.

What has been helpful to you on your rare disease journey?

NORD has been a tremendous support and resource to me and my family, and to CHI. When I first met the dedicated people who work at NORD back in 2011, I felt understood and supported immediately. NORD has helped our family share our story and has helped countless families affected by congenital hyperinsulinism connect and learn about the condition through their information about the disease and sharing out networking opportunities.

NORD has been a huge resource to our organization through its IAMRARE registry platform, and this has helped us to publish in medical journals about what it is like to live with the disease from the perspective of the people who live with it. Patient-powered research helps in the development of new treatments and in developing treatment guidelines.

The annual NORD Summit and Living Rare, Living Stronger Forum have also been enormously helpful, as I have learned so much from other advocates and specialists in rare disease. I have also made so many dear friends by attending them.

I have had the pleasure of planning Rare Disease Day in New Jersey with NORD for a decade and of serving as the Rare Action Network Volunteer Ambassador of New Jersey. In that capacity I worked with NORD on a number of policy issues including a bill to cap co-pays and to establish a rare disease council.

What inspires you about the rare disease community?

It is one of those communities where inspiration is always close by. The people who live with rare disease, who choose to advocate to help others including themselves, are truly incredible. The parents, partners, and other family members and friends who rally around their loved ones and take extra care of them are also so extraordinary. The researchers and physicians who make rare disease the center of their lives simply awe inspiring. Finally, the financial supporters, individual donors and foundations make all this work to improve the lives of people with rare disease possible, so I am so inspired by them, humbled, and truly grateful.