Joslyn Crowe and Laurie Turner of the NNPDF

"At the end of the day, we must go forward with hope and not backward by fear and division."
JESSE JACKSON

Since 1993, National Niemann-Pick Disease Foundation (NNPDF) has served families affected by the different types of Niemann-Pick disease, a severe genetic rare disorder. The organization has worked to build a dynamic and engaged patient support community that embodies the strength, hope, and empathy that all families affected by Niemann-Pick disease deserve. In recent years, NNPDF has enhanced its focus on programs that can support patients and families wherever they are in their journey. This includes emotional support, assistance through a crisis, information about doctors, clinics, and insurance, or just a safe space to share their frustrations and successes.

Niemann-Pick disease is a group of rare, inherited lysosomal storage diseases that affect the body's ability to metabolize fat within cells. Over time, these cells malfunction and can affect organs in the body including the brain, nerves, liver, spleen, bone marrow, and sometimes the lungs. There are several different types of Niemann-Pick, each with varying expectations for symptoms, available treatment options, and impacts on life expectancy. The most commonly recognized forms are Niemann-Pick ASMD (types A and B or Acid Sphingomyelinase Deficiency) and NPC (type C).

Recently, the Niemann-Pick community received the devastating news that the FDA denied the approval of a potential new treatment for NPC. Many rare disease communities have experienced similar situations, feeling they are not heard, and their input has not been given enough credence. Joslyn Crowe, the executive director of the NNPDF since 2018, shares how the community can and must proceed from this point. "*We want to make sure everyone is working toward the same goals and in the same direction. To have everyone united is the strongest chance we have to move forward and achieve our goals. And we must maintain a focus on approval of the therapies we have now, for patient today, not only looking towards future possibilities.*"

Joslyn first learned of the NNPDF through a conversation with a colleague. The organization was going through a period of change and was looking for an executive director. Having worked in patient engagement and stakeholder alliance for more than 15 years and being familiar with the pharmaceutical side of rare disease, Joslyn was interested. She was further inspired by the dedication and commitment of the family-based Board of Directors. Since taking on the position, she has applied her unique range of experience to advance many initiatives at NNPDF with an essential spirit of unity, cohesiveness, and perseverance. Since Joslyn joined, the team and Board of Directors have diversified to include professionals from the pharmaceutical industry, marketing, and a range of different families. She advanced these efforts based on results of a comprehensive community-needs assessment that identified some areas of unmet need in the community – namely, family support services. Joslyn shared, "*while there were other organizations raising money for and dedicated to research, no one was working to provide day-to-day support for families. So, the first thing we did when I came on board was to create a Family Services department*."

Laurie Turner, NNPDF Family Services Manager, had worked previously with a rare disease non-profit for 16 years. She had recently taken a break when she heard about a pilot program that NNPDF was initiating shortly after Joslyn started. NNPDF wanted to build a family services program and had formed a reinvention team to implement many essential changes. Laurie found this opportunity especially compelling and submitted her resume. She was quickly hired and participated in the NNPDF Family Conference on her very first day on the job. She then participated in the four-month pilot study program, learning about the Niemann-Pick families, and assessing their needs. Laurie shared, "*I didn't realize how much I missed being in the rare disease space until I got back into it. It's where I am supposed to be. We have faced some challenges and the road has not always been easy. But our families are amazing, and what they face so bravely every day makes me want to do more for them*." Laurie and the family services program quickly became a go-to resource for people in need, especially during moments of frustration and dashed hope.

While the community has faced many challenges, none were as devastating as the FDA denial of a potential treatment for Niemann-Pick disease type C. It was difficult to consider the best ways to move forward. But that was the commitment at NNPDF. The first step was to host an FDA listening session where the community could present and share their concerns and priorities virtually with FDA reviewers. Joslyn shares, "*the frustration and fear are real, and we are trying to help all of our families. The sense of urgency is paramount right now, and the families are doing what any of us would do if we faced a similar challenge. It is important to recognize that we can still move forward, and there is still potential*." A critical element is the path forward is for the

community to stay united – perhaps more united than ever before. "*If there are any fractures in the community, it is important that we heal them immediately. To be successful we need all 'hands on deck'*." Having a person like Laurie who is active the community and at NNPDF is a critical advantage. She has taken many steps to win the trust and confidence of the families we support. Laurie shares that *"I have had the previous three years within their community, and I am very proud that I am a person they can come to*."

As NNPDF plans for many important developments in 2022, the team will continue to develop new family services programs and remain focused on supporting the effort to bring FDA approved treatment options to Niemann-Pick families. NNPDF will also offer a special fellowship for a young researcher to engage in new initiatives in the Niemann-Pick community and will continue to share new research findings. They will also be working to gather data for an international registry, supporting families through innovative programs, and determinedly working for the Recommended Uniform Screening Panel (RUSP) applications, which would add Niemann-Pick (NPC and ASMD) to newborn screening panels in the US. With endurance, strength, and hope, 2022 will be faced head-on because, as Joslyn shares, "*for this community, it's time, they need it*."