



# Qualitative Study Highlights Need for Increased ASMD Caregiver Support

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Caring for someone with a rare disease, especially a child or a family member, can be a difficult and emotional experience. Many people become a caregiver due to circumstance rather than by choice and lack any formal training. Even for trained professionals, caring for someone living with a rare disease can come with many unique challenges. In many cases, people living with a rare disease require around the clock care and have limited options available for treatment or specialized medical attention. This can increase the emotional and physical toll that caring for someone can impose on a caregiver.

The [National Niemann-Pick Disease Foundation \(NNPDF\)](#) is the leading national organization focused on support for individuals and families affected by any type of Niemann-Pick disease. In recent years, we have worked to expand our focus on meeting the needs of caregivers who support people living with all types of [Niemann-Pick disease](#), including [acid sphingomyelinase deficiency \(ASMD\)](#). In an effort to identify and work to develop more and more targeted support for caregivers in this community, NNPDF recently completed the first-ever study focused specifically on caregiver burden in the ASMD community. The results highlight a critical need for support that can address both the physical and emotional burden that caregivers can experience.

ASMD, also known as Niemann-Pick disease types A, A/B, and B, is a rare, progressive and often life-threatening lysosomal storage disease that currently has no approved treatment options. The progression of disease can vary widely, with some symptoms more easily managed, while people with more progressive forms of the disease require higher levels of support including help with many routine requirements of daily life. The most severe form is rapidly progressive and fatal.

In the study, we conducted a series of one-on-one interviews with caregivers of people living with ASMD to better understand how the disease affects different aspects of their health and life. Of the participants in the study, 100 percent reported that their efforts to provide care for someone living with ASMD have a negative impact to their emotional and financial well-being. The most frequently reported emotional impacts included depression, stress, frustration and concern that the disease symptoms will continue to progress. Participants also reported that their role as a caregiver has an impact on their finances and their concern about how they will address the often significant costs of care. In many cases respondents reported needing to refinance a mortgage and apply for financial assistance. In addition, 86 percent of caregivers in the study reported that they did not receive any formal training on how to provide support and 71 percent were the sole caretaker of a person living with ASMD.

These results help to quantify many anecdotal examples indicating that ASMD caregivers need more and more targeted support and resources to help manage the emotional and financial burden of their role. At NNPfD, we currently offer a number of resources for ASMD families including a guidebook for caregivers of children with rare diseases, information on comprehensive care centers, grief counselling, an emergency hardship program and information on where to find financial and emotional support in local areas. We also host weekly virtual meetings with our family services team where parents and caretakers can share any challenges they might be having and learn about tools that can help. As we work to consider efforts to expand the support network for caregivers, NNPfD is also working to ensure that the experiences of caregivers is taken into account in industry and regulatory efforts to advance research that can lead to a treatment for ASMD.

Another factor that can affect caregivers is the lack of awareness of ASMD, which remains relatively unknown and not widely understood. The lack of information coupled with limited research and no approved treatments can further exacerbate the burden on caregivers. By continuing to raise awareness of the disease, we can help identify more resources and support services that can make a positive difference for caregivers. There is one bright spot on the horizon that could make a profound difference for caregivers. In July 2022, pharma company Sanofi expects to receive a response from the U.S. Food and Drug Administration (FDA) regarding the application for a potential treatment for ASMD. If approved, this will be the first therapy available for ASMD in the U.S. and a sign of hope for the whole Niemann-Pick disease community.

If you are an ASMD caregiver and you need support, please do not hesitate to reach out to the NNPfD family services team at [nnpdf@nnpdf.org](mailto:nnpdf@nnpdf.org). Know that you are not alone in your struggles. We thank you for all the amazing work you do, and we will continue fighting for resources that can improve the lives of everyone impacted by this disease.

To learn more about the study, please visit: <https://nnpdf.org/wp-content/uploads/2022/02/Caregiver-impact-and-burden-of-ASMD-WORLD-02-2022.pdf>

### **About the author**

Joslyn Crowe is the executive director the National Niemann-Pick Disease Foundation (NNPDF), the national patient organization for the Niemann-Pick community. Her work has bridged the nonprofit and pharmaceutical industry realms and includes over 15 years of experience in patient engagement with pharmaceutical companies including Pfizer, Abbott, Novartis, and other on advocacy initiatives and market access strategies.