



Hello!

I hope you all had a summer filled with rest and relaxation. It already feels like forever ago when many of us were together at the NNPDF Family Meeting in Orlando, Florida. It was wonderful to connect with so many families, to see the support for new members of the community and to learn about the innovation happening to help those with NPC. I am including a few photos from our time at the meeting. Thank you to NNPDF for organizing such a special weekend, I hope all of those who attended in person and virtually, found it as rewarding as we did.

During our time in Orlando, we had the good fortune of spending time with Dan and Dee. Dee was diagnosed with NPC when she was in her 40s after many years of unexplained symptoms. She and her husband Dan attended their first NNPDF meeting in 2019 and are very well-known advocates – and friends - in the community. Here they answer some questions about receiving a diagnosis of NPC later in life and give some advice to families who may have similar journeys. You can also read more about their story [here](#).

As always, please reach out to me if you have questions about our work or if there are things you believe we should know.

Kind regards,

Lori

Lori McKenna Gorski
Global Head of Patient Advocacy

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Meet Dan and Dee

Can you tell us a little bit about your lives, and how Dee got diagnosed with NPC?

Dee and I have been married for over 40 years. We have lived in Northern Virginia for 30+ years. Dee was working in the auto insurance industry when we met, and she was sufficiently well respected that she was considered for a VP position within the company. She quit working after our son was born and only worked part time since. I was in the U.S. Army, and we got to spend 5 years in Germany. While there



*Dan and Dee at the 2022 NNPDF
Family Conference in Orlando, FL*

she supported the wives club as a bookkeeper for the annual bazaar. One year they cleared over \$200,000 in a weekend. Of course, she ran the bazaar the next year. After we returned to the states, Dee subbed and volunteered at the schools our son attended, even high school. The year she was the PTSO president, the school was named number one public high school in the country by US News. She was always high performing.

Dee was being seen for migraines at Walter Reed Army Medical Center. In 2005, according to her neurologist's notes, there was a mention of a slight slurring of speech and a slight gait oddity. Her progression with the disease was very slow. She saw various neurologists over the next decade, with continued slow progression, from year to year there wasn't much difference in symptoms. It wasn't until her neurologist from 2005 saw her after a multi-year gap, that the doctors recognized there was a problem. She was initially diagnosed with Spinal Cerebellar Ataxia. They did the screening for

what type and all came back negative. The geneticist at Walter Reed also works at The National Institutes of Health. NIH performed a clinical exome sequencing that came back negative. It was through him that Dee was able to be seen by the Undiagnosed Diseases Program. She was screened in May of 2018, giving 42 vials of blood in 4 days -- 27 in one sitting. She was screened by just about every specialty there is. In August of 2018, we got the call that she had NPC. She had one mutated NPC1 gene, but the screening of lipids and fibroblasts confirmed the diagnosis.

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Meet Dan and Dee cont.

So, we now had a name for the disease. We were told that NPC1 is a fatal disease, with no approved treatment and no cure, and the paperwork we were given said, "...and everyone is dead by age 40." It was at the August 2019 NNPfF Family Support and Medical Conference that we met Dr. Caroline Hastings and our world changed. Dee was enrolled in a clinical trial then and continues to receive infusions every other week at home.

How did it feel to be back at an NNPfF family meeting?

Getting together again with people we had met in 2019 was great. It is important that we see each other – it provides moral support, that we are not alone, and that there are many people and organizations out there that truly care about us. Further, we were able to meet newly diagnosed families. The NPC landscape has changed somewhat for the better since Dee was diagnosed. There are several drugs under development for treatment. There is hope.

Did this meeting feel different at all for you?

I can't really answer this question because we had only been to the 2019 conference. It was different than 2019 but was that because we hadn't gotten together in a couple of years, the stress of COVID lockdowns, or just time passing? I can't tell. At the 2019 conference we were new to the community and, to be honest, still in a bit of shock. It felt good to be able to provide information to others, sharing our journey. We made new friends.

Do you have advice for caregivers of adults with NPC?

My humble advice is to try to show your diagnosed adult how much you love them – even when they don't listen to the things you have repeatedly told them, trying to keep them safe. Tolerate being called a 'nag'. Support them as much as you can. I guess the hardest part is acknowledging that you can't do it all on your own. Reach out to the various resources available, recognizing that those resources vary from state to state, and even county to county or city to city. There is always the NNPfF organization for help in directing your search.



Dan and Dee with other NPC advocacy leaders and the Cyclo Team

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Meet Dan and Dee cont.

What's next for Dan and Dee?

Our adventures continue. In September we have an 11-day bus tour scheduled covering Bavaria and Austria. We will continue to travel as much as we can, while we can. The future with the disease is a complete mystery. When you are so far outside the norm for a disease that is so variable it is difficult to make definitive plans. We know that without a cure, or treatment, Dee is likely to continue to degrade. So we plan for the best, prepare for the worst and try to enjoy each day we have together.



*Dan and Dee at the 2022 NNPDF
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2022 NNPdf Family Support and Medical Conference

It was an honor to participate in the NNPdf Family Support and Medical Conference in conjunction with the INPDA Biennial Meeting. We are excited to continue our collaboration with NNPdf and remain committed to working with the entire community to bring much needed solutions to those living with Niemann-Pick Type C.

