

- Knowing that Niemann-Pick disease type C (NPC) is an ultra-rare, progressive, neurodegenerative disease that occurs in approximately 1:100,000 live births, is heart breaking and living with the fear whether the medication will be approved by FDA is super anxiety inducing. NPC was a late onset and was diagnosed in adulthood in my nephew. This was even harder to accept watching the deterioration and learning the complications of NPC. However, we are grateful for Miglustat and the trial drugs Adrabetadex/Arimoclomol to slow the progression of neurological symptoms associated with NPC. Improvement in swallowing, speech, fine motor skills, and cognition abilities have been noted since my nephew started receiving treatment. Improvements are being tracked via videos and voice recordings. Additionally, measurable improvements are monitored in vision by the ophthalmologist, in speech by the Speech Pathologist and his gate/swallowing abilities by the occupational therapist.
- For my daughter it's frustrating. She realizes that she is losing abilities as time goes on.
- It is extremely painful, depressing and heart-breaking, like living in hell, because medicine is very limited. We see the symptoms of our son with NPC are getting more and more severe but we can do nothing to help him.
- As a caregiver/parent of an adult son with NPC I can tell you that his life has been completely altered by this disease. Those things which he was able to do for himself 5 years ago; drive, work a full time job, manage his finances, eat and function without supervision - are no longer possible. He requires supervision for safe ambulation, safe swallowing, ADLs, and household tasks. His short term memory is all but gone - he cannot remember what he has just eaten, where we have just gone, etc... and requires constant reminders. He is no longer the independant adult he once was and he has no hope of the future he anticipated.
- Devastating, frustrating and depressing. I am 32 years old, diagnosed 4 years ago after my older brother's diagnosis. I have 2 college degrees and worked full time for 4 years before experiencing symptoms. My symptoms were different from my brother's. While he showed physical symptoms along with some early psychiatric issues, mine were solely psychiatric leading to 4 years of misdiagnoses including bi-polar, schizo-affective and schizophrenia culminating in a failed suicide attempt. I have attempted to hold numerous jobs since but have been unable to. My other symptom is related to oral motor in that I have mild dysarthria, coughing while drinking and mild drooling. I have recently begun to expereince some other mild physical symptoms related to balance and fine motor. I contiue to drive but am unsure how long that will be for. I am primarily independent but my parents provide some supervision when necessary when it comes to finances. I have lost all hope of the future I anticipated and no longer believe I will have the job, the house, the family I worked so hard to get. I worry about what will happen if my parents are not around to care for my brother and, ultimately, me.

- My friend tells us about the challenges her family face every day, with ongoing appointments and the unfortunate seizures her daughter has which leads to a lot of time spent at the hospital.
- It is heart rendering for [PATIENT'S NAME], her family and friends to watch her lose her abilities and be in any pain. We all feel helpless as there isn't anything we can do for her.
- Horrendous! Painful, heartbreaking, complete and utter torture watching my child lose her abilities and knowing there is nothing I can do to help.
- Living with someone who has Niemann-Pick type C is upsetting not knowing what is going to happen next. The worry of not only the patient but others around them, the lack of support you receive from doctors who don't know what NPC is.
- The slow neurodegenerative process takes its toll on the patient's life, be it from the ability to communicate, ambulate, thought process and others. It makes it difficult eventually to maintain social contacts with friends and others, as they are aware of your disease and eventual outcome, so they tend to disappear and you're left without friends or people to socially interact with.
- It is extremely distressing observing daily your loved one deteriorate, and there is absolutely nothing you can do to help ease her discomfort and health difficulties. [PATIENT'S NAME] has no downward glare and will often fall over and bruise easily. [PATIENT'S NAME] suffers pains in her hands and legs and walks with a frame and requires a wheelchair when her body is exhausted and the pain is too much for her to be able to walk. [PATIENT'S NAME] "loves" her food, but she is now losing the ability to swallow and has recently had an operation to have a peg fitted. [PATIENT'S NAME] is often sick while she sleeps and this heightens the risk of her choking in the night. Due to this her mother sleeps by her side to ensure she is able to tend to her throughout the night.
- [PATIENT'S NAME] loves school, and they are "brilliant"! But due to the disease [PATIENT'S NAME] is way below her chronological age and will never be able to function the same as her peers. Despite this [PATIENT'S NAME] tries her best and loves to play with all children, but often becomes too tired and experiences pain if she plays for too long. [PATIENT'S NAME] will often become frustrated with herself when her body does not allow her to do the things she enjoys and will voice "I hate being me". This is so distressing to hear and completely heart breaking when you cannot help, change, and/or slow down her rapidly increasing symptoms.
- It is very difficult to live with this disease. It robs you of having a normal life. It robs you of ever having any hope for a normal life. It isolates you. It robs you of your dreams. It is a 24/7 effort to have the best life you can expect.
- NPC is a terrible, degenerative and fatal disease that takes children and other patients piece by piece. Primary decline for most patients is neurological. Something that a child can do today, suddenly they cannot do the same thing tomorrow and once some ability is gone, it is gone forever. This is a horrible life without intervention.
- Constant fear that my son will develop symptoms.

- In all honesty, I was lucky in that NPC only manifested with noticeable symptoms in my senior year of undergrad. I wear two sets of glasses, one for while I am using my laptop at home, and another for just general walking / moving around.
- For my daughter, [PATIENT'S NAME], who just turned 8 on March 3rd, her independence, mobility, and cognition have been affected by NPC; video clips of her at 2 years old show longer statements than she sometimes is able to say today. Her diagnosis of NPC has made her medically-fragile, which has been further complicated by the on-going pandemic; she will be returning to in-person school for the first time in a year and a half, because we have felt compelled to protect her by drastically changing her day-to-day life during the pandemic. When she returns to her second grade class at the local elementary school, a para professional will accompany her everywhere to assist with getting out and holding supplies, eating at lunch, using the restroom, and repositioning into assistive equipment and her power wheelchair. [PATIENT'S NAME] is the light of our family's lives, but NPC has challenged her and us to reimagine what playing outside, dancing together, regular daily self-care, and the future look like. We are blessed to have [PATIENT'S NAME] as our daughter, and appreciate going to receive Adrabetadex twice a month as we have for 5.5 years of her life. [PATIENT'S NAME] diagnosis of early onset infantile NPC has historically caused those patients to pass away before the age of 7; [PATIENT'S NAME] is now the longest living early onset infantile NPC patient to be documented.
- To be different from other normal kids and adults. Being dependent on parents/care givers. So far since 2016 to 2020 I saw gradual progress in my son's gait, speech, swallowing, vertical movement and convergence/divergence of eyesight, and cognition (particularly short term memory). However, after the diagnosis I have been keeping track of his disease progress by taking videos, recording voice and getting measured improvement in vision by OD, improvement in speech, by the Speech Pathologist and OT.
- We received the diagnosis in September 2017, half a year after the birth of our 2nd son. As a result, the disease was also confirmed in our 1st son.

At the moment we lead a "normal" but very conscious family life. Apparently, our children are healthy, not significantly impaired. After the diagnosis, they were prescribed Miglustat. However, we wait daily for changes that may be related to the first symptoms. Every visit to the doctor and every hospital stay, during which examinations are made for symptoms and changes that may be recognisable, is a challenge to our feelings and associated with the hope that nothing will be found.

- Sadness and frustration to a level I have never experienced. With most illnesses there is a level of care that can be experienced, there is a treatment and as such you can have a bit of hope . But with NPC there is nothing we can do but to watch someone we love so much clearly struggle day in day out, it's soul destroying.
- Living with NPC is heartbreaking, traumatic and cruel. It is not only physically challenging it also has negative effects on your mental health. In the 18 months of witnessing my niece suffer with NPC I have seen a variety in symptoms such as; mental frustration, loss of appetite, inability to walk unaided, seizures and hallucinations. On multiple occasions I have seen [PATIENT'S NAME] become angry with herself and even say, "I hate myself", words that should never come from an innocent child's mouth. This is the heartbreak, the trauma and the cruelty this disease causes, an image of my own experiences whilst supporting [PATIENT'S NAME] alongside my elder sister.
- Living with a patient who has NPC completely turns the life of your entire family around. While we were "lucky" enough to be diagnosed early in the disease, everything we do now surrounds our son and his care. Taking him to treatments that are 2.5 hours one way from home has resulted in my inability to work full-time to help support our family. He has now entered 3rd grade and it is becoming harder for him to stay in the classroom with his peers. We think about the future and what we need to do to prepare for his care as he declines. Eventually I will need to be his full time caregiver, we will need to make accommodations at our house etc. Not only have we mourned the loss of our hopes and dreams for our family as a whole but also his dreams and hopes. We spend a lot of time on the phone going over our hospital bills for his treatments.
- It's very hard. I wish no one to feel this way.
- I asked [PATIENT'S NAME] what the most difficult thing about her "differences" are, and she stated that she wishes she could run and dance like other kids. Not being able to keep up with the skillset of other kids around her and therefore being excluded by them, hurt the most. Additionally, she often times gets frustrated about impaired communication (slurring of words making her not always easy to understand) .
- Exhausting, because I always forget a lot quickly. The tremor also upsets me a lot. The swallowing while drinking and when it then runs out of the nose again upsets me very much. The quiet talking, so that hardly anyone understands me in discussions. When walking on stairs I have massive problems. I am exhausted very quickly. I always have to go to the toilet very often. I hardly participate in community life. I am very insecure and this affects my independence.

## What is it like to live with Niemann-Pick type C?

Patient and Professional Statement: Niemann-Pick disease type C

Inpoda

- It is a long and a very hard way to go for children born nearly normal and in good health. It is a frustrating, and a desperate struggle every day to live as their abilities, possibilities and hope of life decline...
- We live all the time in fear for the life of our child. We have no plans for the future. We try to enjoy every day spent together. We don't have time for ourselves. We spend most part of our lives in hospitals.