

Note from Director -

What is Niemann Pick Type C

A simple way of acquainting you with this Lysosomal Storage Disease (LSD) is to compare it to Alzheimer's and ALS (aka Lou Gehrig's- think all of those ice bucket challenge videos in your facebook feed.) Children who received the diagnosis will suffer complete neurological decline; losing the ability to talk, walk, sit, eat and interact with the world around them. Fortunately it is rare, but has been diagnosed far more frequently as an army of parents work on getting the word out. When my son received his diagnosis at the age of two, September 21, 2011, there were thought to be about five hundred cases in the world. Awareness of this disease has led to earlier diagnosis and better supportive care.

It's not contagious, but is "contracted" through some adult activities. (Even if it happens in a test tube I'm going to venture a guess that an adult performed an activity somewhere.) At the moment of conception you inherit half of Mom's chromosomes and half of Dad's to form your own set of 23 pairs of chromosomes. Niemann Pick C (NPC) occurs if both of your parents carried a mutation on one of their Chromosome 18s. Someone is referred to as a carrier of the disease if they have one #18 that doesn't have a mutation and one that does. Recall that you have 23 pairs of chromosomes, only one needs to work (be mutation free) to avoid having the disease. If only one parent passes on the broken 18 then an NPC child would not be born, another carrier would be born. If both parents pass on the broken 18 then the child has Niemann Pick Type C. Twenty Five percent of the time neither mutated gene passes and the new person is neither a carrier or has the disease.

Chromosome 18 has a pretty big job, it is responsible for instructing lysosomes to recycle cholesterol and lipids through the body's cells; not the type of cholesterol in tasty fattening food, the kind the body makes. When the cells get mixed signals from the broken chromosome 18 a traffic jam occurs in the cell and they get overwhelmed with garbage. To complicate the disease there are innumerable different mutation combinations that can occur. A mutation occurs when there is a mix up in the DNA strands, either a letter is added, omitted or reordered, our strands are very very long and the place on the strand that the mutation occurs dictates how fast the disease will onset. However, this is always a fatal

situation. Presently there is no cure or FDA approved treatment for Niemann Pick Type C. Children who are diagnosed very young are not expected to live long lives. In very rare cases the mutations are so benign that the body makes a large enough amount of the NPC1 protein that the lysosomes recycle garbage for many years, not tipping the cells to overwhelmed immediately so the disease takes decades to onset. Commonly it onsets in grade school. Parents with happy, active, beautiful children start noticing bizarre changes in personality and ability. Seizures begin to occur and coordination is lost.

There is a pipeline of drugs and genetic therapies being worked on around the clock. These efforts have been prompted by and funded by families with Niemann Pick Type C and we're simultaneously racing that clock.

Thank you for reading this and letting me do my part to raise awareness of a disease that has altered the course of my life. In a bizarre Kevin Bacon like degree of separation you'll be surprised to learn that you likely know a family that has been touched by this disease. Interestingly it is believed that NPC children cannot contract HIV/AIDS or Ebola because NPC1 protein is needed to recycle the cholesterol that traffic the viruses through the cells. Our community has been collaborating with Ebola researchers in light of the epidemic. If you do know one of these families or catch our awareness campaigns on social media please ask questions, parents of NPC children know the only chance we have is to make connections, someone is going to figure out how to cure this.

With sincerest gratitude for your interest and support,

Shannon Reedy

Director, Mom

Chase the Cure

