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Childhood Disorder

**Niemann Pick Type C Disease: Race to Find a Cure**

In the heart of Oakland, California stands the prestigious Children's Hospital, known for its high quality of pediatric care. Inside, two 6-year-old twins are sharing a hospital bed looking tired and unable to speak. Slowly they are losing everyday bodily functions such as the ability to walk, eat, and play. Soon they will not recognize their parents.

Dressed in matching pajamas, they each hold a stuffed bear while their mom lies between them, an arm wrapped around each girl. The sterile room is filled with nurses, doctors, family, and friends. All are eager and hopeful that the long-awaited procedure the girls are undergoing will save their lives. With an intravenous needle inserted in each girl’s arms there is nothing anyone can do now except wait and hope the medicine works.

The 6-year-old twins, Addison and Cassidy, were both diagnosed with Niemann Pick Type C Disease at the age of two. Niemann Pick Type C Disease, also known as NPC, is a rare genetic disease where large amounts of cholesterol accumulate in the spleen, liver, lungs, and bone marrow, causing damage to the body’s cells. As a result, deterioration of the brain develops. Dr. Marc Patterson, pediatric neurologist at Mayo Clinic in Minnesota, explains in a 2008 podcast titled *Niemann-Pick Disease Type C* that children diagnosed with NPC eventually lose their ability to reason and make decisions. There are also physical disabilities such as balance and coordination that is lost.

Niemann Pick Type C is a disorder related to Lysosomal Disease, a rare metabolic disorder. When babies are born, they are provided genes from both parents. These genes give babies their unique physical appearance as well as how the bodies function inside. Every human is born with the Niemann Pick Type C gene whose job is to regulate cholesterol. When the gene is not working properly it causes a back-up of harmful amounts of cholesterol in the cells which creates NPC.

NPC can hide for months or years before any symptoms occur. Sometimes a stressful illness can trigger the disease. The twins developed mononucleosis as babies that lasted almost one year. The stress from the virus on their bodies caused the Niemann Pick Type C disorder to become active.

NPC is sometimes referred to as the pediatric version of Alzheimer’s disease. Commonly known as an elderly disorder, Alzheimer’s disease can affect children diagnosed with NPC as well as other disorders such as Adrenoleukodystrophy, Alexander disease, Juvenile Huntington’s disease, and Tay Sachs disease. Instead of progressing with age, Addison and Cassidy are regressing the same way a person with Alzheimer’s disease regresses. They no longer can sing their ABC’s and are slowly losing their balance.

Research conducted by the National Niemann-Pick Disease Foundation, Inc. found that symptoms for NPC are not always apparent at birth. They may appear during the first year or not until adulthood. Symptoms usually appear at the ages of 4 through 10. Symptoms include the inability to move the eyes, jaundice, and an enlarged liver and spleen. Not all symptoms appear at the same time. Usually only one or two are apparent in the beginning stages.

NPC is hard to diagnose. Most children are diagnosed with delayed development or a learning disability. It can take several years before the diagnosis of NPC is clarified. There is no known treatment or cure for NPC however there are regimens available to help alleviate the symptoms. In 1997, researchers discovered the location of the Niemann Pick Type C gene on Chromosome 18. This has aided in developing medications used to control seizures and tremors and speech and occupational therapies that help with mobility and functioning on a daily basis. To date, studies have not found anything to cure the disease, but research continues.

Most children diagnosed with NPC do not live to age 20. Doctors informed the parents of Addison and Cassidy that their daughters would not live beyond their teen years. Desperate to fight the disease, the couple turned to various supplements for help, but nothing seemed to work. After more research, it was discovered that Cyclodextrin, an organic sugar compound, is known to break down cholesterol in the body.

Produced from starch, this compound can be found in foods such as butter and salad dressing. It is also found in certain medications and is used in agriculture. Once ingested, the compound has been known to remove cholesterol from cells in certain areas of the body.

The twins’ parents are hopeful that Cyclodextrin will remove the cholesterol build up in their daughters’ cells. After several weeks of adding Cyclodextrin to the girls’ diet, it was determined that the compound needed to be injected into the blood stream to work. After months of working with the U.S. Food and Drug Administration (FDA), they finally received permission to give 2-Hydroxypropyl-beta-Cyclodextrin intravenously to the bloodstreams of the twins.

Addison, Cassidy, and their parents will spend two weeks at The Children's Hospital of Oakland, California where the girls will receive this experimental sugar compound in their blood streams. They are not at the end of their long journey just yet. They have months of weekly treatments to endure before they expect to see results.

In the meantime, the girls’ parents will continue to seek other methods to cure their daughters. While their daughters are receiving the Cyclodextrin, the couple, with the help of medical personnel, will be preparing an application to be submitted to the FDA. They hope to gain approval to intravenously add Cyclodextrin to their daughters’ brains. The process will take awhile but they are determined to find a cure to save Addison and Cassidy.

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