

HI, MY NAME IS ABBY



AGE 7

Motto: "Shining a Light on NPC1"

Diagnosis: Niemann Pick Type C1

High School Team: Hyde Park Cheer

Abby was born premature and spent 59 days in the NICU. She was diagnosed with Niemann Pick Type C1 (NPC1) after her older sister, Belle, was also diagnosed in Spring 2016. NPC1 is a rare genetic mutation. There are only 500 known cases in the world, with just 100 of them in the United States. Patients with NPC1 lack functioning NPC1 proteins, which is responsible for the trafficking of cholesterol. Because cholesterol cannot be trafficked, it accumulates in all of the cells in the body.

Patients with NPC1 first experience a cognitive decline, similar to that in Alzheimer's disease. Abby is currently receiving an experimental treatment called VTS 270. Because the drug does not cross the blood brain barrier, treatment is administered via lumbar puncture every two weeks at Dell Children's Hospital.

Despite all of her challenges, Abby is very outgoing and makes friends everywhere she goes. She loves to dance, sing, and perform. She loves animals, particularly dogs. Her older sister, Belle, is a sustaining child in the CC4C family.