HI, MY NAME IS ABBY

I am 6 years old and have been diagnosed with Neimann Pick type C1

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

Patients 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.

Despite all of her challenges, Abby is very outgoing and makes friends everywhere she goes. She loves to dance and sing. She loves to perform and be the center of attention. She loves animals, particular dogs. She is supported by her school, and her older sister is a sustaining child of CC4C.

CC4C

CHEYANNA'S CHAMPIONS 4 CHILDREN

Uplifting the Rare and Undiagnosed

Abby's Motto:

"Shining a Light on NPC1"