

HI, MY NAME IS KYLE

I am 10 years old and undiagnosed.



When Kyle was an infant he was developmentally delayed, had seizures, and high muscle tone. His muscles were so tight it was difficult to put him in a car seat. The doctor's he saw decided his issues were due to his premature birth and he began physical therapy to help him with the tightness. His milestones were delayed, but he was determined and always found a way to get what he wanted. When he couldn't crawl he learned to get where he needed to go by rolling.

When he was able to get up on his arms, he learned to crawl by dragging his legs behind him. As he got stronger with therapy he eventually began walking. Almost as soon as he started walking he began showing signs of pain in his legs. He would scream out but didn't have the words to tell us what was wrong. As he got older, he was able to tell us the muscles in his legs and ankles hurt and that pain happens any time he plays hard. Track and Field day at school last year left him in so much pain that it was hard to get out of bed the next day. Kyle also suffers from stomach pains, severe dyslexia and anxiety.

The geneticist ordered whole exome sequencing on Kyle to compare his DNA to his twin sister's in a search for a diagnosis for her medical issues. Much to our surprise, Kyle and his sister were diagnosed with an ultra-rare genetic condition called ADNP Syndrome.

ADNP is a gene in the brain that can affect 400 different systems in the body. To date, only 210 children in the world have been diagnosed, and Kyle and his sister are the only two with their particular mutation. Kyle works very hard in school and loves to play with his friends. He is learning to recognize when he feels anxious and talk through his feelings. He is learning to listen to his body as well, when it tells him he needs to take a break. Since before he was able to talk he has been signing and spreading joy to everyone around him, and most of all he never lets anything stop him.



Kyle's Motto:

“Always Do Your Best”