# Rowan's Story

After a year of looking, we finally received a diagnosis for Rowan that explains his walking delay. Genetic testing revealed that he has a very rare progressive motor neuron disease called spastic paraplegia type 4, or SPG4. It is a type of hereditary spastic paraplegia (HSP). There is no cure or treatment for SPG4, only physical therapy to strengthen the muscles and procedures/medications to manage the spasticity.

Typically, SPG4 symptoms don't present until later in life and progress rather rapidly. Early onset of the disease often means a slower progression, so we are hopeful his condition will remain stable for some time. However we can't know what Rowan's future looks like, as HSP is a "heterogenous" disease, which means that every person with SPG4 can suffer different symptoms, progression and severity.

Learning that Rowan has SPG4 was devastating. This was the call we had been dreading, and we are just starting to process the shock and come to terms with the diagnosis. We're learning everything we can about HSP and SPG4, becoming better informed about disability, and continuing to take Rowan to physical therapy to strengthen his muscles and give him the best range of motion and most independent mobility he can achieve.

The future of this disease is uncertain, which is scary - but it's also hopeful. There's no one way that SPG4 progresses, so we are going to hold expectations high for Rowan. No matter what, he is so deeply loved by all of us, and he is the absolute happiest and sweetest child I've ever known. He is creative, outgoing, determined, and joyful. And of course he has us to advocate for him, care for him, and help support him whichever way he needs us to as he grows.

These are some other details about Rowan:

- Rowan is two and a half. Born Feb 2022.
- Diagnosed with torticollis in infancy but resolved with treatment.
- Rowan met milestones at unusual times (ex. army crawled before sitting independently)
- At 15 months Rowan could not stand or walk independently. He had been cruising on furniture for 5+ months, but never progressed past that milestone.
- PT did not suspect anything genetic until watching how slow Rowan's progress was over time. Referred him to neuro and genetics, suspecting cerebral palsy.
- Had a clear MRI scan and clear muscular dystrophies genetic panel. Six months later, his geneticist/neurologist decided to run a genetic panel for diseases similar to CP.
- Diagnosed with SPG4 per that genetic panel in May 2024. He has received one round of Botox for spasticity, but that wore off quickly.
- Rowan was also diagnosed with a speech delay at 15 months, but as of now he has nearly caught up to peers.
- I have been tested for SPG4 and have been cleared. Waiting for my husband to be tested. We anticipate Rowan's condition is de novo however.
- Rowan has one older sister, age 4, who has not shown any signs of SPG4.
- Currently Rowan has uncomplicated SPG4, but obviously only time will tell if he has a more complicated form of the disease.
- Rowan crawls, uses a walker, and is learning how to use a wheelchair to get around. He is active, wiggly, highly social and the happiest little guy ever!