

## **Maddon's Story**

When Maddon was 2 ½ years old, he began seeing a speech therapist for speech delays. One day his speech therapist mentioned she thought it would be a good idea to have one of her physical therapists take a look at Maddon, as he walked mostly on his tippy toes. Up until that point we had always thought how Maddon walked was cute, he even earned the nickname 'twinkle toes.' After we saw the PT, they suggested we go to Seattle Children's Hospital to do further evaluation. Initially they had thought Maddon had CP or autism. Once we ruled those out, we started connecting some dots that potentially his paternal grandmother's undiagnosed disability might be something hereditary. It would take over 6 years of testing (blood tests, MRI's, nerve conduction studies, and many physical assessments) until we were finally able to get a positive diagnosis of HSP SPG 4.

Even with a diagnosis, it has been difficult to explain to people what Maddon has needed and what his limitations are since so few people are familiar with HSP. Maddon's symptoms primarily include weakness in his legs, spasticity in his calves, gait abnormality, toe-drop, and overall balance and tripping issues (although he is independently mobile). A typical day for Maddon since our first visit at Children's has involved trying to walk in many different types of AFOs, although we never have found ones that work well for him; many appointments with neurologists and physical therapists; home PT and stretching; and various trial and errors with medications such as muscle relaxers – all with little to no improvement on his symptoms.

With Maddon's symptoms he has not been able to play sports, participate fully in PE, or be a part of many outside of school activities such as birthday parties with his peers, which has been very hard for Maddon emotionally. Going to school overall has been difficult as Maddon trips and falls at least a few times a day, making us worry that he may get really injured with a bad fall – not to mention how it impacts him emotionally when other kids notice him walking differently and falling at times. Simple scenarios like being on stage for a class holiday performance have been hard, with the risk of a fall being even more worrisome.

Now Maddon is in his first year of middle school and doing well despite his challenges. He has a few close friends and really excels in computers and video gaming. We continue to see doctors at Seattle Children's and work on maintaining his mobility and range of motion with physical therapy and stretching at home. To date Maddon has been very lucky that his symptoms have not progressed, and we are hopeful that will continue. Our family has also started getting involved in raising awareness and funds for the SP Foundation in the past few years since Maddon's diagnosis. We have found this to be a really positive way to do something productive as a family given the reality of the situation. We've had tons of fun putting on these events – including raffles at the local pizza restaurant, golf and poker tournaments and a private wine tasting dinner. We are hopeful that with continued awareness and fundraising there will be a cure for Maddon.