Austin Nace's Story

Austin was born in June of 2005, a big boy of 10 lbs. 5 oz.! Diagnosed as having had very common newborn pulmonary hypertension, he spent one week in the NICU on oxygen and then we brought him home having no idea that there was anything more to worry about.

After a year, however, he was not hitting milestones, had constant respiratory issues, ear infections, and had a curve to his spine. Our small town pediatrician in rural NY had read an article years before, and it triggered her to send Austin for x-rays. We got the call (yes, someone thought it was a good idea to tell me this over the phone) a week before his 1st birthday, "He looks to have a genetic disease called Mucopolysaccharidosis, (known as MPS) and you need to get him to a geneticist as soon as possible." If you look it up on the internet, which is of course what we immediately did, you will see that it is terrifying information. There we sat together in middle of the night, reading how our sweet boy would have dwarfism, painful malformed joints throughout his body, spinal cord compression, heart damage, airway obstruction, vision loss, hearing loss, and would suffer progressive brain damage. We read the words "10 year average life expectancy", and "1 in 120,000 births". These kids, because of the disease, look more like each other than they do their siblings. We saw the pictures… we recognized the look. Within a few weeks he had a geneticist, and he was officially diagnosed with MPS Type 1- Hurler's Syndrome, the most aggressive form of the disease.

There is no cure for any of the forms of MPS, but there are two treatment options: transplant (bone marrow or stem cell), or ERT (enzyme replacement therapy). We had no long term data as to the effectiveness of ERT, because it had been approved less than two years before his diagnosis. After gathering all the information we could, we went the route of ERT; which he still receives to this day and will continue to receive once a week as long as he is with us. After 5 years of 1-2 hr drives to the children's hospital for his 5 hour IV infusions we were finally able to switch to home infusions! As of writing this we figured that he has had 485 of these infusion days, having only missed one time for the swine flu when he was a toddler.

So far, Austin has had 2 Lifeport placement surgeries, adenoid removal, carpal tunnel release in both hands – as well as all 8 fingers trigger released, two hernia repairs (both which failed within 2 months), as well as sedated MRI's of his brain, spine and neck. He regularly sees Metabolism, Orthopedics, Cardiology, Pulmonology, Nephrology, ENT, and he will soon see Dental, Ophthalmology. All of these specialists are at Children's Hospital of Philadelphia (CHOP). He also has physical therapy 3x week, speech therapy and occupational therapy 2x week.

What none of the internet medical journals or physicians could tell us is how amazing this boy would be despite his disease. They couldn't teach us like Austin could that life with Hurler's would be so hard, but that life doesn't need to be easy to be good. That you can hurt and struggle and still laugh and share joy. That every lousy trip to the hospital can be an adventure where you get to make "new best friends" and care for caregivers. Austin loves life, he loves people, and he changes everyone who knows him for the better. We are humbled by his strength and faith, and we are so grateful to the good people at the Shannon Daley Memorial Fund for giving us the opportunity for us to share him with you!

For more info on MPS – www.mpssociety.org