Jonah's Just Begun Holiday Remit Drive & 7th Annual Newsletter

November 2017

Dear Supporters:

It's been a hard year for so many of our friends and families who have lost loved ones or been uprooted during the numerous natural disasters and murderous violence. I feel deeply for these people. I know that acute feeling of loss and despair.

In February of 2010, we took Jonah in for an MRI of the brain at our pediatrician's request. Jonah had a big head, no biggie, we all thought nothing of it. Jonah was 22 months old at the time of diagnosis and meeting all his milestones; Jonah was better than normal, he was the cutest baby in all of Brooklyn. Jeremy had randomly had the day off that day, as one of the actors had a family emergency and filming was shut down for the day. This never happens. In retrospect, I couldn't imagine what I would have done if he hadn't come.

Our neurologist examined Jonah from head to toe. An intern stood motionless in the corner. I couldn't take my eyes off of her. She knew something. Our doctor asked a lot of questions, she was thorough. Jonah put his shirt back on and started to play with Dad in the corner. I sat down with the doctor; she braced herself and told me our son had a form of mucopolysaccharidosis (MPS). As she started to tell me about the different forms, the tears just came. I looked at Jeremy. He was bouncing a ball in the corner with Jonah. Panic swept over his face. He came closer. I asked the doctor: "Are you telling me that our son is going to die?"

I don't remember her response exactly. She did her best to calm me, as the facts were not all in. We needed to see the geneticist and do a blood test to confirm which MPS. She opened up a drawer and had a stack of prescriptions all written out for us. Eye exam; full body X-ray, another MRI, ENT, GI, sleep study and the geneticist. One by one we went to all of these appointments. Each one found supporting symptoms. Six weeks later we were called back in to see our geneticist. Jeremy and I hoped that this was just a misunderstanding - a lab mistake. We clung to that idea. We had arrived at the same time as our geneticists. She looked at us standing at the elevator and said: "I'll take the stairs."

She told us that Jonah had Sanfilippo type C. The most rare form, but possibly more attenuated; there are 4 versions of Sanfilippo A-D. There was next to nothing written about type C, so she was basing things on the literature from type A papers. When I asked her if this was a death sentence, she just sat there for so long. She finally said: **"There are treatments today never dreamed possible just 5 years ago. This does not have to be a death sentence."**

Once home we put Jonah down for a nap and we silently wept. In that first week after diagnosis, we filed for our non-profit status, reached out to scientists and started planning our first fundraiser.

On a shoe-string budget, we have brought this disease to the brink of a clinical trial.

We have gotten this far on sheer will. Donations have been coming in, in dribs and drabs and our fundraisers are never enough. Luckily, what JJB lacks in funding we make up in charm. We have been very fortunate to capture the pro-bono services of professionals in this space that have saved JJB from going belly up. This year when the research invoices came due it was our sister foundations that picked up the tab**. Seven years into this quest we are now confident that we have a drug bound for clinical trial**!

"When we dream alone, it is only a dream. When we dream together it is the beginning of a new reality." - Friedensreich Hundertwasser

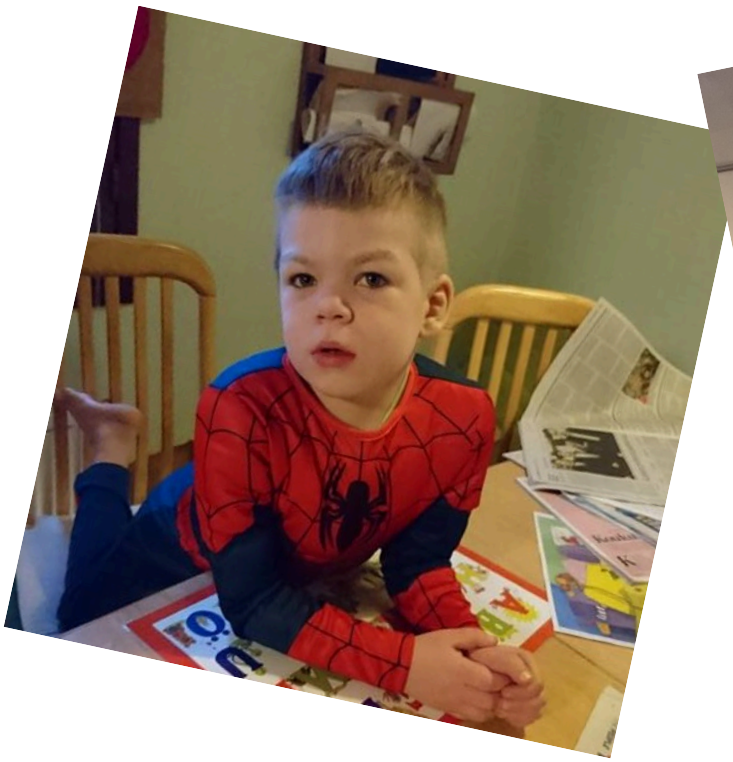
Still, at the mercy of big pharma, JJB and HANDS have been able to scrape together enough dollars, euros, and pounds to cover the cost of our gene therapy research. We need a partner to manufacture and commercialize our treatment. Like a broken record, over and over we hear: "Your disease is too rare. Your disease is too rare. Your disease is too rare."

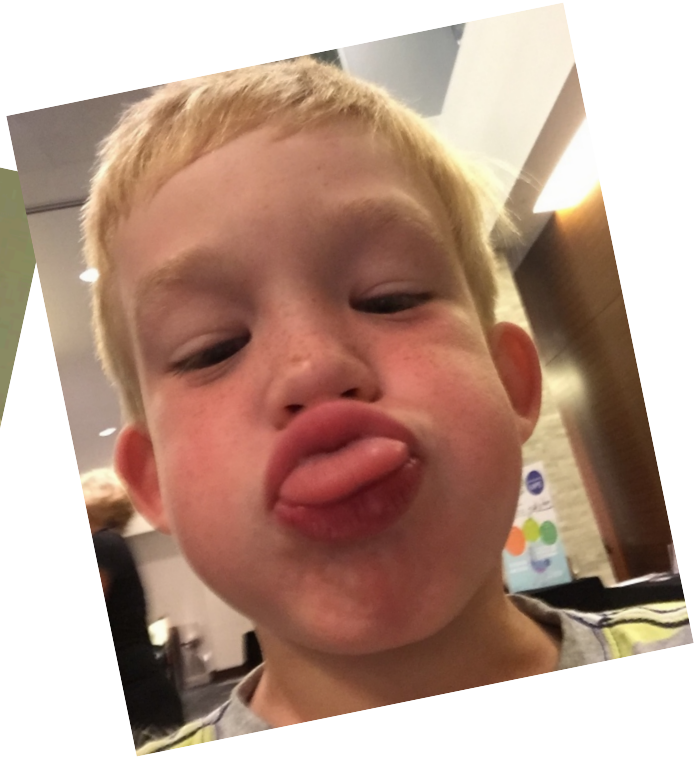
Potential partners have added a second verse: "What are you doing about finding more patients?"

Finding a needle in the haystack... How do we help doctors diagnose a rare disease that they know nothing about? There are 7,000 rare diseases. Maybe they read about Mucopolysaccharidosis in med school but they have never seen a patient in real life....or have they? Sanfilippo children are almost always diagnosed as autistic, which is the catch-all for "I don't know what's going on with this child, but they are cognitively delayed, so we will go with autism". Sanfilippo children can go anywhere from years to decades living with the wrong diagnosis.

Here's the thing...

Sanfilippo children have distinctive facial features. Medically termed "coarse". Stand two blue-eyed blonde Sanfilippo boys together, roughly the same age and they could be brothers. Spiderman lives in Finland, he's 6 in this picture Jonah is 8 here. Jonah was taking a selfie, but his silly face highlights the coarse features of a Sanfilippo child. In layman's terms notice the boy's prominent foreheads, wide fleshy eyelids, low nasal bridge with wide fleshy tip, full lips, low detached fleshy earlobes and unmanageable hair. A Sanfilippo child's hair is so thick and kinky it does what it wants to do.





My point... These kids are out there they are just misdiagnosed or undiagnosed. If a doctor saw these pictures then they'd be able to recognize the next Sanfilippo child that walked into their office. No matter color of hair or skin all of our kids have these distinctive features.

This past year JJB spent its entire fortune, made up mostly of energy, perfecting and marketing three programs:

ConnectMPS: Our patient registry. Here families can upload diagnostic information, answer survey questions and make themselves accounted for. The goal is to help academics learn more about the MPS's and encourage pharma to create treatments for us. **https://connect.patientcrossroads.org/?org=ConnectMPS**

Mission: Hide and Help (MHH): an awareness campaign. Think social experiment. WE need YOU to help us make this viral (FYI Sanfilippo syndrome is not contagious.) The inspiration for MHH comes from finding the rare. Like being able to find a rare Pokemon card, it makes you cool to do so. It makes you a HERO to find and diagnose a child with a rare genetic defect.
https://missionhideandhelp.org/

Face2Gene: A facial recognition app designed to aid doctors in diagnosis, based on facial features. FDNA works with the same facial recognition software that Facebook uses. Our Sanfilippo community has helped train the FDNA software to capture Sanfilippo by uploading hundreds of pictures of our children.
http://suite.face2gene.com/

Our goal… Diagnose and tag that gorgeous face, then get that mug on record.

TAG-DIAGNOSE-RECORD equals saving lives.

Early diagnosis leads to treatments. Sanfilippo syndrome does not have Newborn Screening because there is not a treatment. The rationale is why tell parents that their baby has a terminal illness if they don't have a treatment available. The ignorance is bliss philosophy. JJB is here to tell you otherwise. **We can't have treatments without a diagnosis.**

JJB is calling on you to help save lives. Sadly, there is no glossy newsletter this year, overflowing with positive energy and pretty pictures. JJB is pinching every penny and saving all our energy for a clinical trial**. Donate today and we will make sure that your donation lives on forever in a cure.**

Sincerely,

Jill Wood

Jill Wood

Co-Founder and Treasurer