AS HEALTHY AS POSSIBLE

June 2026, Newsletter

Welcome to our newsletter!

We want to connect our community more and make more information and resources available to you. Find us at AsHealthyAsPossible.net and please contact us with feedback at: info@ashealthyaspossible.net

FUNDRAISING

All donations made this year will be matched to reach our **$120000 goal**. Your $100 donation becomes a $200 donation!

JOHN'S STORY

Our youngest of three boys was born in 2018. We were thrilled to welcome him into our family. Based on our experience with our older two, we started to suspect some delays before he was a year old. Our pediatrician put us in touch with the local Childhood Developmental Services Agency and we began therapies and a search for a diagnosis. In January of 2020 we discovered through exome sequencing that he had a rare mutation to his gene responsible for POLR2A. While we scrambled to figure out the implications and impact of this diagnosis we plunged into the COVID pandemic.

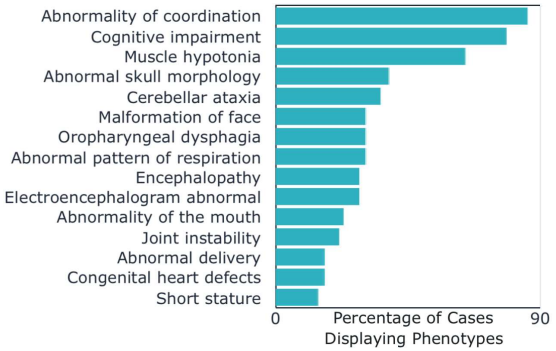
While we understood how lucky our family was to have an early diagnosis; there was no clear path on how to proceed forward and what to expect. The first year after the diagnosis was emotional and stressful for our entire family to maintain the daily expectations of what a family "should" be like, while trying to learn about entirely new topics to us. While the pandemic brought forms of isolation to everyone, we also felt isolated from other families based on the needs our family was developing and the changes we needed to make to meet those needs.

A doctor early on told us that in cases of rare disease it is often those dealing with it directly that become experts, more so than the doctors. We've found this to be true. It is a tremendous balancing act, but I believe our family has been blessed in many ways beyond what we as parents would be able to provide ourselves that has allowed our family to thrive. We continue on in our journey navigating rare disease and we still have a lot to learn. Becoming a part of this community has been wonderful for us, and created stronger family bonds between all of us. We are so happy John is a part of our family and shares his joy with us.

REGISTRY UPDATES

A POLR2A focused patient registry is being hosted at the University of Pittsburgh using the Geneial platform to securely record patient surveys about children and individuals with a POLR2A mutation. The goal for this registry is to understand where patients might be similar or different and how different POLR2A mutations might relate to disease symptoms or severity. We would urge everyone to participate and fill out surveys if you or your child has a genetic testing document indicating a POLR2 mutation. Currently, 56 people have expressed interest, 27 have started to fill out information, 24 have indicated genetic testing results, and 6 have completed health surveys.

Aggregated and anonymous results from the surveys are available to participants and are updated as more participants join (a sample is shown below). We are hoping for 20-25 more participants by the end of the summer. To join, please contact POLR2AR@pitt.edu or visit https://advocate.geneial.com/prescreen/polr2a



COMING SOON

A POLR2A research initiative is starting up at Mayo Clinic, the University of Wisconsin, and the University of Pittsburgh. We will be sharing more about this as development progresses.

NORD® National Organization for Rare Disorders

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