FOUNDATION FOR

HAO-FOUNTAIN SYNDROME

USP7 GENE

HAO-FOUNTAIN SYNDROME

VISION

Providing a future of possibilities for those who are diagnosed with Hao-Fountain Syndrome.

MISSION

Our mission is to cure Hao-Fountain Syndrome. We do this by funding research and identifying more patients.

USP7

CONTACT US

If your child has been diagnosed with Hao-Fountain Syndrome, we would love to connect with you and provide you with as much information about the genetic disorder as we can. While this is a rare diagnosis, you are not alone!

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WHO WE ARE

We are a registered 501(c)(3) non-profit organization dedicated to those who are affected by a mutation of the USP7 gene. A mutation of USP7 causes a neurodevelopmental disorder called Hao-Fountain Syndrome. Those affected are often developmentally delayed, have white matter abnormalities, speech impairment, are diagnosed with Autism Spectrum Disorder and more.

WHAT IS HAO-FOUNTAIN SYNDROME

USP7 Gene

USP7 is a protein-coding gene that plays a role in tumor suppression, transcriptional regulation, immune response, and endosomal protein recycling. Individuals who are born with a mutation in USP7 have been found to have a neurodevelopmental disorder.

Gene Mutations

Mutations in USP7 are either point mutations or gene deletions. Mutations are diagnosed through either whole exome sequencing or chromosome microarray analysis. The inheritance pattern of the disease caused by USP7 mutations is autosomal dominant, which means that someone who receives a single copy of an abnormal USP7 gene from either parent may have this disorder.

Symptoms

- Developmental Delay/ Intellectual Disability
- Speech Impairment
- Autism spectrum disorder Neonatal hypotonia
- Significant feeding problems Hypogonadism
- Eye abnormalities (strabismus, myopia, nystagmus, or other) Reflux/Gerd
- Abnormal brain MRI
- Hypotonia
- Contractures
- Short stature
- Difficulty gaining weight Chronic constipation
- Chronic diarrhea
- Seizures
- Abnormal gait
- Aggressive behavior