

# GET IN TOUCH NOW

**Website** : [usp7.org](http://usp7.org)

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



Individuals who are born with a mutation of the USP7 gene have been found to have a neurodevelopmental disorder called Hao-Fountain Syndrome (HAFOUS).

Mutations 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. To date most cases are de novo, meaning the mutation/deletion has been spontaneous and not inherited.



## FOUNDATION FOR HAO-FOUNTAIN SYNDROME

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



# ABOUT US

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

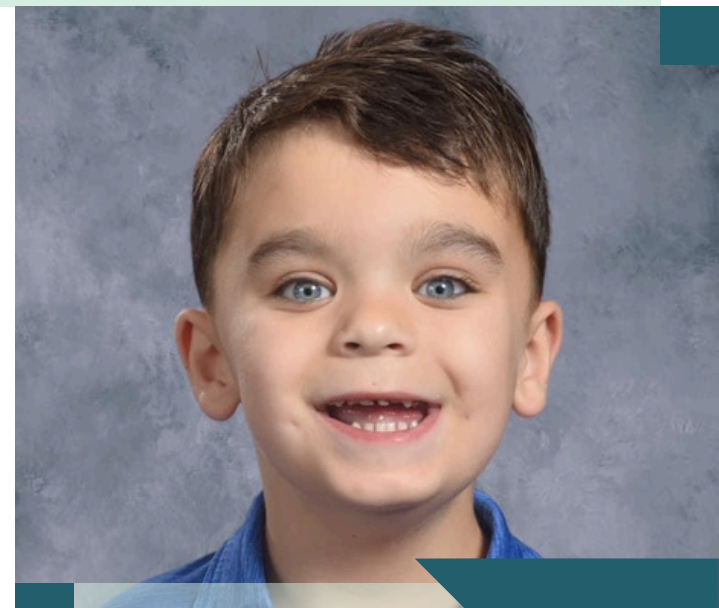
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# SIGNS AND SYMPTOMS\*

## Physical Characteristics

- Dysmorphic Facial Features
- Abnormal MRI
- Hypotonia
- Eye Anomalies (esotropia, myopia, strabismus, nystagmus)
- Feeding Problems
- GERD
- Seizures
- Neonatal Hypotonia
- Hypogonadism
- Asthma
- Abnormal Gait
- Difficulty Gaining Weight
- Sleep Apnea/Sleep Disturbance
- Chronic Constipation
- Short Stature
- Scoliosis or Kyphosis
- Neonatal Poor Suck
- Contractures
- Small Hands
- Small Feet
- Excessive Weight Gain
- Chronic Diarrhea
- Decreased Fetal Movement
- Hip Dysplasia
- Hearing Difficulties



## Behavioral Characteristics

- Speech Delay (some nonverbal)
- Developmental Delay/Intellectual Disability
- Behavioral Anomalies (aggressive behavior, temper tantrums, impulsivity, compulsivity, stubbornness, manipulative behavior)
- Autism Spectrum Disorder
- ADHD
- Skin Picking

\*Hao-Fountain Syndrome has a wide range of physical and behavioral characteristics. It is important to note that not all patients display all symptoms and characteristics. Symptoms can vary from mild to severe.