## Vision Statement

Providing a future of possibilities for those who are diagnosed with a mutation of USP7.

## Mission Statement

Our mission is to cure USP7-related diseases. We do this by funding research and identifying more patients.

## Publications

USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder

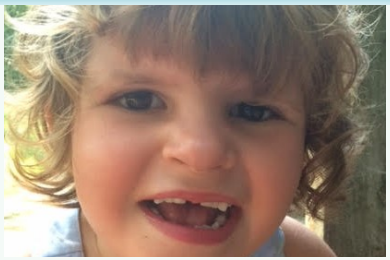
- Hao, Fountain, et al.
  Molecular Cell 59(6) Sept 2015

**Foundation for USP7-Related Diseases**

**www.usp7.org**

# USP7-Related Diseases Physician Brochure

FOUNDATION USP7 DELETION OR MUTATION



## 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.

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.

## 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

## Suggested Tests

Once diagnosed, there are certain tests that are recommended for each patient. These tests include:

1. Measurement of IGF-1 and IGF-BP3 to screen for growth hormone deficiency
2. A brain MRI after 40 months of age to assess for abnormalities of white matter
3. Full assessment by a speech pathologist
4. Full assessment for physical and occupational therapy
5. Formal cognitive and behavioral testing by a licensed pediatric psychiatrist
6. A sleep apnea test/sleep study
7. An EEG test to test for abnormal electric activity that could cause/ predispose seizures
8. A consultation with a gastroenterologist for any reflux, vomiting, or chronic constipation/ diarrhea issues
9. An assessment by a pediatric ophthalmologist