COHEN SYNDROME OVERVIEW

WE ARE ON THIS JOURNEY TOGETHER.

The Cohen Syndrome Research Foundation (CSRF) was started by parents of children with Cohen Syndrome. CSRF is a resource for families looking for information, a repository for medical professionals seeking standards of care and research studies, and a driving force behind researchers seeking to improve outcomes of those with Cohen Syndrome. Our mission is to improve the lives of Cohen Syndrome individuals by supporting and advancing research.

Genetics

Cohen Syndrome is a rare genetic disorder that is caused by variants in the VPS13B gene. Variants in the VPS13B gene are believed to inhibit the production of the functional VPS13B protein. Researchers are still studying how the VPS13B protein affects development in individuals with Cohen Syndrome.

Cohen Syndrome is an autosomal recessive disorder, which means that both parents must carry and pass on an abnormal copy of the gene in order for a child to be affected. When both parents are carriers, there is a 25% chance of Cohen Syndrome with each pregnancy. Cohen Syndrome is formally diagnosed through Whole Exome Sequencing (WES).

Symptoms

The symptoms of Cohen Syndrome are variable and there is no way to predict what symptoms will be present in each child. Common symptoms include:

- Failure to thrive in infancy
- Developmental delay
- Intellectual disability
- Autism or autistic-like traits
- Neutropenia (low levels of white blood cells leading to frequent infections)
- Retinitis Pigmentosa (a degenerative disease of the retina leading to vision loss)
- Myopia (nearsightedness)
- Microcephaly (small head size)
- Hypotonia (weak muscle tone)
- Distinctive facial features including thick hair and eyebrows, long eyelashes, wave shaped eyes, a bulbous nose, prominent upper central teeth, narrow hands and feet, slender fingers
- Truncal obesity in late childhood or adolescence
- A cheerful and friendly disposition

Please click here for a more comprehensive explanation of symptoms.

Therapies

Early intervention and supportive therapies are extremely valuable. These services may include:

- Physical therapy
- Occupational therapy
- Feeding therapy
- Speech therapy – many children with Cohen Syndrome are nonverbal or have delayed speech and might benefit from Augmentative and Alternative Communication (AAC)
- Behavioral therapy – consult a pediatrician to determine if an autism assessment is right for your child.

Medical Specialists

Currently, there is no cure for Cohen Syndrome. Here are some recommendations for medical providers that may be able to assist on your journey (please note that CSRF is not run by medical professionals, and this document should not be treated as medical advice):

- Hematology - A hematologist can help monitor neutrophil levels to mitigate the risk of serious infection. Congenital neutropenia can be treated with granulocyte-colony stimulating factor (G-CSF), which stimulates the bone marrow to produce infection-fighting neutrophils. Many patients receive regular G-CSF injections to reduce the effects of neutropenia.
- Ophthalmology – While there is no cure for the vision loss associated with Cohen Syndrome, eyeglasses can help with myopia.
- Gastroenterology - Many individuals have difficulty gaining weight in infancy and experience feeding difficulties during their lifetime.
- Developmental Pediatrics
- Orthopedics
- Endocrinology
- Anesthesia - Anesthesia use in Cohen Syndrome patients should be carefully monitored and always delivered in a hospital setting. Please request the anesthesia protocol for more information.

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The Cohen Syndrome Research Foundation is a 501(c)(3) nonprofit organization.