Inheritest®
CARRIER SCREEN

Cohen syndrome

What is Cohen syndrome?

Cohen syndrome is an inherited disease with variable signs and symptoms. Cohen syndrome is known to be caused by mutations in the gene that produces the VPS13B protein; however, the function of the VPS13B protein is currently unknown.[1]

What are the symptoms of Cohen syndrome and what treatment is available?

The symptoms of Cohen syndrome are variable and may include[1,2]:

- Developmental delay
- Intellectual disability
- Microcephaly (small head)
- Hypotonia (poor muscle tone)
- Nearsightedness
- Retinal dystrophy (degeneration of the retina of the eye)
- Joint hypermobility (large range of movement)
- Neutropenia (abnormal white blood cell count)
- Overly friendly behavior
- Truncal obesity
- Narrow hands and feet
- Delayed puberty
- Distinctive facial features

Treatment is supportive and focuses on prevention of complications and management of symptoms.[2]

How is Cohen syndrome inherited?

Cohen syndrome is an autosomal recessive disease caused by mutations in the *VPS13B* gene.[1] An individual who inherits one copy of a *VPS13B* gene mutation is a carrier. Carriers are not affected with Cohen syndrome. An individual who inherits two *VPS13B* mutations, one from each parent, is expected to be affected with Cohen syndrome.

If both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Cohen syndrome?

Cohen syndrome is a rare condition with an unknown prevalence. It may be seen more commonly among the Finnish population.[3]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/cohen-syndrome

Cohen Syndrome Association: http://cohen-syndrome.org/

National Organization for Rare Disorders (NORD): http://rarediseases.org/rare-diseases/cohen-syndrome/

References

1. Cohen syndrome. *Genetics Home Reference*. Available at http://ghr.nlm.nih.gov/condition/cohen-syndrome Accessed February 15, 2016.
2. Cohen Syndrome. *National Organization for Rare Disorders*. Available at http://rarediseases.org/rare-diseases/cohen-syndrome/ Accessed February 15,2016.
3. Kolehmainen J, *et al*. Cohen syndrome is caused by mutations in a novel gene, COH1, encoding a transmembrane protein with a presumed role in vesicle-mediated sorting and intracellular protein transport. *Am J Hum Genet*. 2003;72:1359-1369