Genetics Uncoded:

Facts about Cohen Syndrome

What Your Test Results Mean

Carriers show no symptoms of Cohen syndrome and are not at risk to develop Cohen syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Cohen Syndrome Explained

Cohen syndrome is an inherited disorder characterized by developmental delay, intellectual disability, small head size, and weak muscle tone. Other features include progressive nearsightedness, degeneration of the light-sensitive tissue at the back of the eye, hypermobility, neutropenia, and distinctive facial features. Management of Cohen syndrome is supportive.

How the Genetics Work

Cohen syndrome is an autosomal recessive disorder caused by variants in the *VPS13B* gene. In general, individuals have two copies of the *VPS13B* gene. Carriers of Cohen syndrome have a single variant in one copy of the *VPS13B* gene, while individuals with Cohen syndrome have variants in both copies of the *VPS13B* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

Clarity Genetics, LLC
5231 NW 33rd Ave
Ft. Lauderdale, FL 33309

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