

Cohen Syndrome

What Your Results Mean

Test results indicate that you are a carrier of Cohen syndrome. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of Cohen syndrome for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.



Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for Cohen syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Cohen Syndrome Explained

What is Cohen Syndrome?

Cohen syndrome is an inherited disorder characterized by developmental delay, mild-to-moderate intellectual disability, small head size (microcephaly), and weak muscle tone. Other symptoms may include progressive nearsightedness, degeneration of the light-sensitive tissue at the back of the eye, hypermobility, neutropenia (low levels of white blood cells) with frequent infections, and distinctive facial features. Affected individuals may also present with obesity and an overly friendly disposition.



Prognosis

Prognosis is considered unfavorable; however, treatment can improve an individual's quality of life. The effect on lifespan is not well-defined.

Treatment

Treatment of Cohen syndrome is symptomatic as there is no cure. Addressing symptoms as soon as possible through early intervention, physical therapy, occupational therapy, and speech therapy can be beneficial.



Resources

Cohen Syndrome Association

<http://www.cohensyndrome.org/>

National Organization for Rare Disorders (NORD)

<https://rarediseases.org/rare-diseases/cohen-syndrome/>

National Society of Genetic Counselors

<https://www.nsgc.org/>