SeSAME Syndrome, Enlarged Vestibular Aqueduct, Digenic

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of SeSAME syndrome. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of SeSAME syndrome, Pendred syndrome or *FOXI1*-related hearing loss for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. This is a condition that has digenic inheritance-mutations in two different genes result in varying symptoms. 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 are carriers for SeSAME syndrome, *FOXI1*-related hearing loss, or Pendred syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

SeSAME Syndrome, Enlarged Vestibular Aqueduct, Digenic Explained

What is SeSAME Syndrome, Enlarged Vestibular Aqueduct, Digenic?

SeSAME syndrome is characterized by seizures, sensorineural deafness, ataxia (lack of muscle coordination), intellectual disability, and electrolyte imbalance (low levels of potassium and magnesium in the blood, hypokalemia and hypomagnesemia, and metabolic alkalosis). It is also known as EAST syndrome for symptoms of epilepsy, ataxia, sensorineural deafness, and tubulopathy (kidney problems in structures known as tubules). Seizures tend to start in early childhood and are typically of the generalized tonic-clonic seizure type (also known as grand mal seizures), but the seizures usually respond well to medication. Non-progressive cerebellar ataxia and hearing loss start later. The ataxia is likely the most debilitating feature of the syndrome.

SeSAME syndrome is due to mutations in the *KCNJ10* gene. Mutations in this gene can also be causative of Pendred syndrome. Pendred syndrome is an inherited condition that affects inner ear formation and the thyroid gland. Because individuals with Pendred syndrome are born with varying degrees of inner ear malformation, they are born with or soon develop moderate to profound deafness. Some individuals also have difficulty with balance. Affected individuals also have enlarged thyroid glands, and though this does not usually lead to thyroid malfunction, the enlargement (goiter) may lead to discomfort and difficulty swallowing and breathing.

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Prognosis

Prognosis of SeSAME depends on the severity of signs and symptoms. Prognosis of Pendred syndrome is generally good. Most individuals with Pendred syndrome present with profound deafness before they are able to speak, but a minority develop progressive hearing impairment later on. Some individuals will develop hypothyroidism and 60% of individuals develop goiters. However, thyroid problems can be managed with nutritional iodine. Pendred syndrome does not affect life expectancy.

Treatment

Treatment of SeSEAM syndrome includes antiepileptic medication to address seizures, physical, educational and speech therapy, hearing aid and management of the kidney and electrolytes problems.

Treatment for Pendred syndrome is focused on addressing hearing loss. Children may be fitted for hearing aids early in life. Educational programs for the hearing impaired and cochlear implantation may also be considered. For goiters, treatment may include radioactive iodine and/or surgery.

Resources
National Association of the Deaf
https://www.nad.org/
Epilepsy Foundation
https://www.epilepsy.com/
National Society of Genetic Counselors
https://www.nsgc.org/

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