What Your Test Results Mean

Testing results indicate that you are a carrier of Pendred syndrome. Carriers typically show no symptoms of Pendred syndrome; however, carriers are at an increased risk of having a child with Pendred syndrome. Risk for the current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Pendred Syndrome Explained

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. There is no cure for Pendred syndrome; however, some families opt for the use of hearing aids or cochlear implants.

How the Genetics Work

Pendred syndrome is an autosomal recessive disorder of hearing loss caused by mutations in the SLC26A4 gene. In general, individuals have two copies of the SLC26A4 gene. Carriers of Pendred syndrome have a single mutation in one copy of the SLC26A4 gene while individuals with Pendred syndrome have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with Pendred syndrome is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.