



Genetics Uncoded:

Facts about Pendred Syndrome



What Your Test Results Mean

Carriers typically show no symptoms of Pendred syndrome; however, carriers are at an increased risk of having a child with Pendred syndrome. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● 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 variants in the *SLC26A4* gene. In general, individuals have two copies of the *SLC26A4* gene. Carriers of Pendred syndrome have a single variant in one copy of the *SLC26A4* gene while individuals with Pendred syndrome have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with Pendred syndrome 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.