

# Pendred Syndrome

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## What Your Results Mean

Test results indicate that you are a carrier of Pendred syndrome. Carriers typically show no symptoms. Risk for 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.

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 Pendred syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

## Pendred Syndrome Explained

### What is 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.



### Prognosis

Prognosis 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 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

**American Society for Deaf Children**

<http://deafchildren.org>

**Hearing Loss Association of America**

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

**National Association of the Deaf**

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

**National Society of Genetic Counselors**

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