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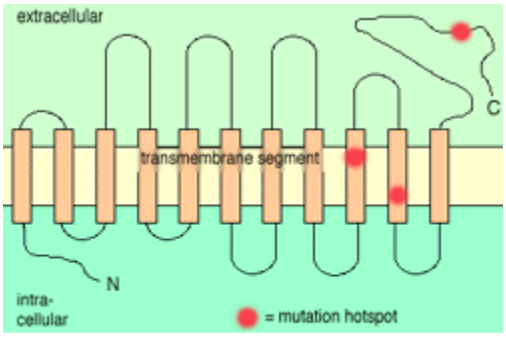
Pendred syndrome



Model of the human pendrin protein, based on the predicted amino acid sequence. The approximate positions of mutations in some pendred syndrome patients are shown in red.

Pendred syndrome is an inherited disorder that accounts for as much as 10% of hereditary deafness. Patients usually also suffer from thyroid goiter. The recent discovery of the gene for Pendred syndrome illuminates a disorder that has confounded scientists for more than a century.

In December of 1997, scientists at NIH's National Human Genome Research Institute used the physical map of human chromosome 7 to help identify an altered gene, *PDS*, thought to cause pendred syndrome. The normal gene makes a protein, called pendrin, that is found at significant levels only in the thyroid and is closely related to a number of sulfate transporters. When the gene for this protein is mutated, the person carrying it will exhibit the symptoms of Pendred syndrome.

Because goiter is not always found in Pendred syndrome patients, it is possible that a defective pendrin gene will turn out to be responsible for some cases of deafness that had not previously been attributed to this disorder. The discovery of pendrin should also stimulate new angles of research into thyroid physiology and the role of altered sulfur transport in human disease.

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