

Pendred Syndrome

Also known as: Goiter-Deafness Syndrome

Clinical Characteristics

In addition to hearing loss, Pendred syndrome is characterized by thyroid goiter. While most (about 75%) of individuals have normal thyroid function despite the enlargement of the gland, some are clinically diagnosed with hypothyroidism. About 40% of those affected develop goiter in late childhood; the rest usually do so in early adulthood. Some individuals may have balance problems due to inner ear abnormalities.

Pendred Syndrome and Hearing Loss

Hearing loss is bilateral, sensorineural, and generally severe to profound. The loss is usually congenital and non-progressive, though mild-to-moderate progressive hearing loss has occurred. All individuals with Pendred syndrome have temporal bone abnormalities. Vestibular dysfunction is common; over 60% of individuals have bilateral dilation of the vestibular aqueducts. This can occur with or without cochlear hypoplasia.

Natural History

Pendred syndrome is diagnosed in both males and females, and in all ethnicities. The prevalence of the syndrome is unknown, though some estimates place it between 1 in 10,000 and 1 in 100,000. It is also estimated that Pendred syndrome accounts for 7-10% of cases of congenital deafness. Pendred syndrome is the most common form of syndromic deafness.

Hearing loss is usually present at birth. Thyroid abnormalities, including goiter, generally do not appear until late childhood or early adulthood. The appearance and severity of symptoms is variable, even among affected family members. The prognosis for those diagnosed with Pendred syndrome is good, overall, if the symptoms are well-managed. Intelligence is usually normal, though many individuals require early speech therapy and educational intervention to account for hearing loss. Life expectancy is normal.

Genetics

The characteristics of Pendred syndrome are secondary to a mutation in the gene *SLC26A4*, which is found on the long arm of chromosome 7 and makes a protein functional in the thyroid and inner ear called pendrin. The alteration of this protein leads to defective iodide transport and organification. The exact mutation is identified in about 50% of cases. Pendred syndrome is **autosomal recessive**. This means that both parents of an affected individual must be carriers for the condition. Whenever two carriers for Pendred syndrome have a child, there is a 1 in 4 (25%) chance the child will

be affected, a 2 in 4 (50%) chance the child will be an unaffected carrier just like their parents, and a 1 in 4 (25%) chance the child will not be affected nor a carrier. Overall, there is a 75% chance with each pregnancy the child will **not** have Pendred syndrome. . If one parent has Pendred syndrome, the chance of having an affected child depends upon the carrier status of the other parent. If both parents have Pendred syndrome, every single child born to that couple will also have the syndrome.

Management

The diagnosis of Pendred syndrome is made clinically. Genetic testing is available for confirmation of diagnosis, and carrier testing and prenatal diagnosis if the mutation in the family is identified.

An initial audiologic evaluation of an individual with Pendred syndrome should consist of assessment of auditory acuity (ABR emission testing, pure tone audiometry), computed topography of the temporal bones to look for abnormalities, and vestibular function studies. Audiometry should be repeated every three to six months initially to check for signs of hearing loss progression. Hearing aids should be used as early as possible, and cochlear implants can be considered for those with severe to profound hearing loss. Speech therapy and sign language communication are appropriate interventions.

In addition, semiannual or annual examination by an endocrinologist is indicated. A first visit should include a perchlorate discharge test and thyroid function tests. Individuals with thyroid dysfunction should be treated accordingly.

A scheduled appointment with a geneticist and/or genetic counselor is recommended.

Some physicians suggest that activities such as weightlifting and contact sports should be avoided in order to prevent an increase in intracranial pressure for those with dilation of the vestibular aqueduct. The schools and parents should work together to create an Individual Education Plan which addresses a child's specific learning needs.

Resources for Families

Statewide Genetics Program

Phone: 608-267-7148

Fax: 608-267-3824

Email: meyeram@dhfs.state.wi.us

Wisconsin First Step Hotline

Phone: 1-800-642-7837 voice/TTY

Website: www.mch-hotlines.org

Wisconsin Bureau for Deaf and Hard of Hearing

Phone: 1-608-266-3118 voice/TTY

Website: www.dhfs.state.wi.us/sensory

Regional Children and Youth with Special Health Care Needs Centers

Centers in Green Bay, Wausau, Milwaukee, Madison, and Chippewa Falls

Website: http://dfhs.wisconsin.gov/DPH_BFCH/cshcn/index.HTM

Parent-to-Parent of Wisconsin

Phone: 1-888-266-0028

Email: mathea@shsmh.org