Waardenburg Syndrome

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Waardenburg syndrome is an inherited genetic disorder which may cause hearing loss and partial albinism. It is believed to account for 2-3% of cases of congenital deafness (deaf at birth). It affects both males and females, and people of all ethnic backgrounds. It is estimated to occur in 1 of 42,000 individuals.

Symptoms and types

Waardenburg syndrome is divided into four types, based on symptoms. Individuals are considered to have Waardenburg syndrome type 1 if they have 2 major or 1 major plus 2 minor criteria from the lists below.

Major criteria:

- Born deaf or hard of hearing (congenital sensorineural hearing loss) (58% of individuals)
- Brilliant sapphire blue eyes or two different color eyes
- White lock of hair on the forehead
- Immediate family member with Waardenburg syndrome
- Inner corner of the eye displaced to the side (dystopia canthorum)

Minor criteria:

- Patches of light or white skin
- Eyebrows extending toward middle of face
- Nose abnormalities
- Premature graying of the hair (by age 30)

Waardenburg syndrome type 2 is defined as having all the features of type 1 except dystopia canthorum. 77% of individuals with type 2 have hearing loss.

Waardenburg syndrome type 3, or Klein-Waardenburg syndrome, is similar to type 1 but includes muscle contractures. Waardenburg syndrome type 4, or Waardenburg-Shah syndrome, is also similar to type 1 but includes Hirschsprung's disease (a gastrointestinal malformation).

Diagnosis

Babies born with Waardenburg syndrome may have the characteristic hair and skin changes and hearing loss. However, if the symptoms are mild, Waardenburg syndrome may go undiagnosed until a family member is diagnosed and all family members are examined. Formal hearing tests can be used to detect hearing loss.

Treatment

Different symptoms of Waardenburg syndrome appear in different people, even within the same family. Some individuals will require no treatment, while other may need surgery or eye or other abnormalities. No special diet or activity restrictions are needed, and Waardenburg syndrome does not usually affect the mind.

Genetic counseling

Because of the way Waardenburg syndrome is inherited (autosomal dominant) in types 1 and 2, an affected individual has a 50% chance in each pregnancy of having an affected child. Since symptoms can vary, there is no way to predict whether an affected child will have milder or more severe symptoms than his/her parent. Inheritance of types 3 and 4 is more complex, but genetic counseling can help assess the risk of passing Waardenburg syndrome on to a child.

Information for this article was taken from:

Bason, L. (2002). Waardenburg syndrome. eMedicine, accessed at http://www.emedicine.com/ped/topic2422.htm