Waardenburg syndrome

Genetic Disorders Project Waardenburg Syndrome Brian Tokay March 4, 2011 Waardenburg Syndrome is an inherited disorder characterized by different levels of hearing loss and changes in hair and skin pigmentation. A common trait of Waardenburg Syndrome is different colored eyes or very bright blue eyes, a low hairline, connected eyebrows, some may have a patch of white hair or grey hair by the age of twelve, and a wide space between the eyes.

There are at least four types of Waardenburg Syndrome-Types 1, 2, 3, and 4 with type 1 and 2 being the most common. Physical characteristics identify with what type of Waardenburg a person has. For example, wide set eyes are common in type one and hearing impairments occur in twenty percent of type one Waardenburg. Hair discoloration is common in type two and hearing loss occurs in fifty percent of type two Waardenburg.

Type three is called Klein-Waardenburg and is similar to type one but includes muscle problems, and type four is called Waardenburg-Shah which is also similar to type one but includes intestinal problems. People may have only some traits of the syndrome and some may have all traits. Rare cases associate Waardenburg Syndrome with other problems like intestinal disorders and spinal problems, and cleft lip/palate which is a facial deformity.

Waardenburg Syndrome was discovered by a Dutch doctor named Petrus Johannes Waardenburg who noticed that people with different colored eyes often had hearing impairments so that made him go on to study over one thousand people in deaf families and found that some had physical characteristics in common such as the wide set eyes or low hairline. Doctor

Waardenburg later discovered that the cause of the syndrome is that it is passed from parent to child just as blood type and hair color is passed, and because

Waardenburg Syndrome is a dominant condition, a child usually inherits it from one parent who has a malfunctioning gene. There is a fifty-fifty chance that the child of someone with Waardenburg Syndrome will also have the syndrome. Treatments for Waardenburg Syndrome depend on the type of syndrome someone has. Usually no treatment is necessary unless abnormalities require surgery. Genetic counseling can help determine the risk of passing the syndrome on to a child, and attention should be focused on the hearing deficits and hearing aids should be used.

If a person has intestinal or muscle/skeletal problems are present then treatment may be required for them. People with type three or four Waardenburg Syndrome may have additional problems which can lead to complications such as severe constipation, complete deafness, self esteem issues, and a slight risk for a muscle tumor called rhabdomyosarcoma. If these issues are present then treatment will be required such as laxatives, counseling, surgery, and education to learn sign language and possibly special education classes.

Once hearing problems are corrected or the person adjusts to hearing loss, people with the condition should lead a normal life. There are no shorter life expectancies for people with Waardenburg Syndrome as to people without it. Counseling may be required for people with self esteem issues regarding the syndrome and medication may be required for the secondary problems

related to the syndrome, but it usually does not interfere with someone’s life as a whole. Waardenburg Syndrome