Usher Syndrome Type 1B (MYO7A)

Usher syndrome is a condition that involves various degrees of deafness and gradual impairment of vision. Those with Usher syndrome type 1 are usually born deaf, and begin to lose vision while still children. A number of different genetic mutations can cause Usher syndrome type 1, including mutations in the MYO7A gene (Usher syndrome type 1B). MYO7A encodes for the protein myosin VIIA, which is involved in molecular transport. MYO7A is produced in the retina and inner ear, and is important for proper functioning. In the inner ear, it is involved in the production and maintenance of the hair-like stereocilia, which are essential for hearing. The vision loss from Usher's syndrome is due to the condition retinitis pigmentosa, which involves the gradual deterioration of retinal rod photoreceptor cells (leading to night blindness), followed by cone receptor cells (leading eventually to complete blindness). In addition to deafness, Usher syndrome type 1 affects the ability to balance. Children with the condition are typically slow to stand up and walk.

Usher syndrome type 1 affects overly 12,000 people in the USA alone. Roughly half of these are due to MYO7A mutations. Usher syndrome type 1 is more common in certain ethnic groups such as Ashkenazi Jews and the Acadians (Cajuns) of Louisiana. The condition is autosomal recessive, which typically requires both parents to be carriers, and usually are asymptomatic.

Sources

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