CLRN1 Gene

Subjects: Genetics & Heredity
Contributor: Vicky Zhou

clarin 1

genes

1. Normal Function

The *CLRN1* gene provides information for making a protein called clarin 1. This protein is probably involved in normal hearing and vision. Clarin 1 has been found in several areas of the body, including sensory cells in the inner ear called hair cells. These cells help transmit sound and motion signals to the brain. This protein is also active in the retina, which is the light-sensing tissue that lines the back of the eye. Although the function of clarin 1 has not been determined, studies suggest that it plays a role in communication between nerve cells (neurons) in the inner ear and in the retina. Clarin 1 may be important for the development and function of synapses, which are junctions between neurons where cell-to-cell communication occurs.

2. Health Conditions Related to Genetic Changes

2.1. Usher Syndrome

At least 15 mutations in the *CLRN1* gene have been identified in people with Usher syndrome type III, which is characterized by a combination of hearing loss and vision loss. Some affected individuals also have problems with balance and coordination. *CLRN1* gene mutations cause a form of the condition known as Usher syndrome type IIIA (USH3A). This form of Usher syndrome is rare in most countries, although it represents about 40 percent of all Usher syndrome cases in the Finnish population.

Several *CLRN1* gene mutations change single protein building blocks (amino acids) in the clarin 1 protein. In some cases, these mutations lead to the production of an abnormally short version of the protein or prevent the production of any functional clarin 1. Other mutations insert or delete small amounts of DNA in the *CLRN1* gene, which probably impairs the normal function of the protein. It is unclear how a missing or altered clarin 1 protein leads to the signs and symptoms of Usher syndrome type IIIA.

Two particular *CLRN1* gene mutations are most common in families of Finnish ancestry. One mutation, sometimes called Finmajor and written as Tyr176Ter or Y176X, leads to the production of an abnormally short, nonfunctional version of clarin 1. The other mutation, written as Met120Lys or M120K and also known as Finminor, substitutes

the amino acid lysine for the amino acid methionine at protein position 120. This mutation appears to disrupt the protein's normal function.

2.2. Retinitis Pigmentosa

Retinitis pigmentosa

3. Other Names for This Gene

- USH3
- USH3A
- USH3A_HUMAN
- Usher syndrome 3A
- Usher syndrome type 3 protein

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