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My46 Trait Profile

Usher syndrome

Usher syndrome is characterized by hearing loss and progressive vision loss. There are three different types of Usher syndrome that result in different ages of onset and severity of hearing and vision loss. It can be caused by mutations in any one of many different genes.

Characteristics of Usher syndrome

There are three types of Usher syndrome. In all three types, affected individuals have hearing loss as well as Retinitis Pigmentosa (RP). RP causes slowly progressive vision loss, specifically difficulty with night vision and blind spots in the peripheral (side) vision. Vision loss can progress to "tunnel vision", that is, loss of peripheral vision resulting in a decreasing field of vision. Most individuals with Usher syndrome retain some degree of central vision.

Individuals with Usher syndrome type 1 are often born deaf or are found to have severe hearing loss in the first year of life. Onset of RP is typically first noticed in childhood. Individuals with Usher syndrome type 1 also have problems with balance; children often start walking late, around 18 months to 2 years old.

Individuals with Usher syndrome type 2 typically have mild to severe hearing loss that is usually detected in childhood. Onset of RP usually begins as a teenager or adult.

Individuals with Usher syndrome type 3 have hearing loss often beginning in childhood and becoming progressively worse into adulthood. Onset of RP usually begins as a child or teenager. Some individuals with Usher syndrome type 3 may have difficulty with balance.

Diagnosis/Testing

Clinical diagnosis of Usher syndrome is based on tests performed by an ophthalmologist and audiologist. In an ophthalmology clinic, testing often involves examination and photography of the back of the eye, visual field tests to determine the field of vision and identify any blind spots, and an electroretinogram (ERG). And ERG measures how well the light sensitive cells at the back of the eye are functioning. Testing in an audiology clinic often involves a hearing test to measure how loud sounds need to be before a person can hear them.

Most individuals with Usher syndrome type 1 have changes or mutations in one of six genes: MYO7A, CDH23, PCDH15, USH1C, USH1G, and CIB2. Most individuals with Usher syndrome type 2 have mutations in one of three genes: USH2A, GPR98, DFNB31. Most individuals with Usher syndrome type 3 have mutations in one of two genes: CLRN1 and HARS.

Management/Surveillance

Management of Usher syndrome includes the use of hearing aids or cochlear implants, low vision aids, speech therapy, and sign language training, as needed. Some individuals with Usher syndrome may develop cataracts, which can be surgically removed.

Mode of inheritance

Usher syndrome is inherited in an autosomal recessive pattern. This means that an individual has to inherit mutations in a gene associated with Usher syndrome (i.e., one from each parent) to be affected with Usher syndrome. If both parents are carriers of a mutation in the same gene associated with Usher syndrome, they have a 1 in 4 (25%)

chance with each pregnancy of having a child with Usher syndrome.

Risk to family members

Parents of a child with Usher syndrome are carriers of Usher syndrome. If a sibling of a child with Usher syndrome is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of Usher syndrome.

Special considerations

None

Resources

Genetics Home Reference: Usher syndrome
http://ghr.nlm.nih.gov/condition/usher-syndrome
National Institute on Deafness and Other Communication Disorders: Usher syndrome
https://www.nidcd.nih.gov/health/hearing/pages/usher.aspx
The Coalition for Usher Syndrome Research
http://www.usher-syndrome.org/
The Foundation Fighting Blindness
http://blindness.org
The Helen Keller National Center
http://www.hknc.org
Hear See Hope
http://www.hearseehope.com/

References

Keats BJB, Lentz J. Usher Syndrome Type I. 1999 Dec 10 [Updated 2013 Jun 20]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2014. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1265/

Keats BJB, Lentz J. Usher Syndrome Type II. 1999 Dec 10 [Updated 2013 Aug 29]. In: Pagon RA, Adam MP, Ardinger HH, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2014. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1341/

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