Synonyms: Graefe-Sjögren syndrome, Graefe-Usher syndrome and von Graefe’s syndrome

The syndrome is a genetic defect causing retinitis pigmentosa and congenital deafness. There may also be vestibular dysfunction. Deafness is usually congenital but loss of visual acuity and visual fields, progressing to complete blindness, occurs in the teens and 20s in both Usher's syndrome type 1 (USH1) and Usher's syndrome type 2 (USH2).\[1\]

Classification

Usher's syndrome is divided into 3 types, although a 4th type has been suggested. The 3 basic types are called USH1, USH2 and USH3 but further subdivisions use letters such as USH1a and USH1b.

A certain amount of gene mapping has been done but not for all types.

Epidemiology

The incidence is about 1 in 25,000. About 3-6% of hearing impaired children have the condition. Types 1 and 2 are more common than type 3 and together account for 90-95% of Usher's syndrome and about 10% of all children born deaf. Usher's syndrome represents about half of all people who are both deaf and blind. Most cases of type 3 come from Finland.

Genetics

It is autosomal recessive and so both parents must be carriers or even affected, although spontaneous mutation can occur. USH4, if it is a separate entity, is X-linked. Most authorities do not accept type 4 and it is said that there are 3 types with 8 different genes implicated.\[2\]

The relevant gene locus varies between types. Even the chromosome varies. The chromosomes that bear the mutation for the variations USH1, USH2 and USH3 are numbers 11, 1 and 3 respectively, although some of the subgroups are on different chromosomes.

Presentation

- Usher's syndrome type 1 (USH1) produces profound deafness from birth and there are severe problems with balance. Hearing aids offer little or no benefit and most communicate by sign language. Poor balance makes them slow to sit without support and they rarely learn to walk before the age of 18 months. At first, sight seems normal but they usually have some problems with vision by the time they are aged 10. Night vision is affected first but it progresses rapidly until they are completely blind.
- Children with Usher's syndrome type 2 (USH2) are born with moderate-to-severe hearing impairment and normal balance. The degree of hearing loss varies but most attend normal schools and benefit from hearing aids. They communicate with speech and often lip read. The higher frequencies are more affected than the lower ones. The visual problems in USH2 tend to progress more slowly than in USH1. USH2 causes visual field defects that appear in the early 20s.
- Children with Usher's syndrome type 3 (USH3) have normal hearing and normal or mildly impaired balance. Hearing deterioration starts around puberty. Blind spots appear in the late teens or early 20s. By middle age they are usually blind.

Visual loss is progressive but auditory loss is constant,\[3\] at least in USH2.

Loss of vision\[4\] usually starts with night blindness and this is followed by loss of peripheral vision. Some degree of tunnel vision can continue until quite late.

In USH1 there is impairment of the vestibular system\[5\] that accounts for the lack of balance. It is normal in USH2 but visual feedback contributes to balance. The adequacy of vestibular function in USH3 is unknown.

Usher's syndrome can lead to and present with psychiatric disturbances which can be difficult both to investigate and to manage.\[6, 7\]
Differential diagnosis

Deafness and retinitis pigmentosa are rarely found together. Most people who have retinitis pigmentosa and hearing loss probably have Usher's syndrome type 1 (USH1) or Usher's syndrome type 2 (USH2).

In the early stages it may be thought of purely as an auditory problem and the problem of sight is not anticipated unless there is a family history. About 1 in 4 with retinitis pigmentosa has Usher's syndrome.

The other major cause of deafness and blindness is congenital rubella.

Investigations

Electronystagmography (ENG) to detect vestibular problems and electroretinography (ERG) to detect retinitis pigmentosa aid early detection.

Management

Early diagnosis[8] of Usher’s syndrome is important to permit special educational training to facilitate coping with the combined hearing and visual loss. The programme will depend on the severity of the auditory and visual impairments as well as the age and abilities of the individual.

They will benefit from:

- Adjustment and career counselling
- Access to technology such as hearing aids, assistive listening devices or cochlear implants[9] (which appear to enhance the quality of life)[10]
- Orientation and mobility training
- Communication services and independent living training that may include learning Braille, low vision services, or auditory training

Prevention

Although much work has been done on genetic research and gene mapping,[11] gene testing and testing for the carrier state is not yet available. There is no ante-natal diagnosis. If a child is born with the condition the risk of an affected sibling is 1 in 4.

History

Charles Howard Usher was a Scottish ophthalmologist who was born in Edinburgh in 1865. He trained at Cambridge and St Thomas’ Hospital. He described the syndrome in 1914 in a work called On the inheritance of retinitis pigmentosa, with notes of cases.[12] In it he reviewed the syndrome, describing the pathology and inheritance of 69 cases.

He was appointed ophthalmic surgeon to the Aberdeen Hospital for Sick Children and also worked in the Aberdeen Royal Infirmary. Apart from military service in Salonika during the First World War, he remained in these posts until he retired in 1926. He died in 1942.

Further reading & references

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