Usher syndrome: an overview

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Usher syndrome is the most common hereditary condition that affects both vision and hearing. It is an autosomal recessive condition characterised by retinitis pigmentosa (RP) and bilateral sensorineural hearing loss [1,2].

Usher syndrome is the cause of approximately 10% of all hereditary deafness, with deafness existing at birth or developing soon afterward [1]. The diagnosis of Usher syndrome is generally made by ophthalmoscopic examination showing RP in a congenitally deaf patient [3]. As the typical funduscopic findings of RP are not usually evident until late in the first decade, diagnosis is often delayed until late in the development of the disease [3]. These ophthalmic symptoms are summarised in the table below but characteristically cause night blindness. People with RP may develop other treatable eye diseases, such as glaucoma, cataract and macular cystic changes [4].

Key ophthalmic symptoms in Usher syndrome
▪ Night blindness
▪ Blind spots resulting in loss of peripheral vision
▪ Impaired colour vision

There are three types of Usher syndrome that are differentiated by the severity of hearing loss, presence / absence of balance problems and age of onset [5]. The three types are:

- Type 1: Born with severe hearing loss, progressive vision loss caused by RP apparent in childhood and balance affected.
- Type 2: Born with severe hearing loss, progressive vision loss caused by RP begins in adolescence and balance not affected. This is the most common type.
- Type 3: Normal hearing at birth: hearing and vision loss starts in adolescence and balance may (but not always) be affected.

The typical funduscopic findings of RP consist a triad of signs: a pale optic disc, attenuated blood vessels and scattered areas of pigment clumping initially seen near the equator [3]. Alongside examining the retina, visual fields should always be assessed to determine the extent of peripheral visual loss. Colour vision should also be assessed as 63% of patients with RP develop acquired colour vision defects, usually tritanopia [6].

Furthermore, electrophysiological studies including electroretinography (ERG), electro-oculography and dark adaptation studies are useful in ensuring early diagnosis and to determine the extent of RP. A subnormal dark adapted b-wave of the ERG indicating diminished rod acitivity is often the earliest ocular abnormality detected in RP [3]. Optical coherence tomography (OCT) may also be useful to determine if there are macular cystic changes. Any patient diagnosed with retinitis pigmentosa should be referred to ENT for an evaluation of auditory acuity and vestibular (balance) function, given that 10% of these patients will present with hearing loss [3].

Currently, there is no specific treatment for patients with Usher syndrome or RP, but genetic studies of RP are essential in finding a cure or prevention [1]. The treatment of Usher syndrome involves a multidisciplinary approach and is symptomatic and supportive, involving use of low vision aids, hearing aids and initiating genetic counselling. Despite no cure for RP, research is currently being undertaken to determine the benefits of daily vitamin A supplementation to slow the progression of retinal degeneration. Due to the long-term adverse effects of vitamin A (e.g. liver disease) more results from larger randomised controlled trials need to be published prior to this being nationally recommended.

In conclusion, early diagnosis of Usher's syndrome is crucial in helping affected patients manage their situation. A screening programme which enables congenitally deaf patients to have an early ophthalmic consultation may be beneficial in identifying Usher syndrome earlier and thus reducing the impacts RP on a patient's life.

References

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