

Is there a link between Usher syndrome type II and balance problems or vertigo?

Information Monitoring Summary

Documentary research

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Notice to readers

The information in the following pages is not intended to be an exhaustive review of the literature. The goal was to make directly relevant selected information more readily available. Accordingly, not all articles or documents dealing with the topic have been reviewed.

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SUMMARY

Usher syndrome type II is characterized by moderate to severe congenital deafness, normal vestibular function, and retinitis pigmentosa usually appearing in the patient's late twenties or early thirties. Clinicians in the deaf-blindness program run jointly by the Institut Nazareth et Louis-Braille and the Institut Raymond-Dewar have noticed that certain patients with this form of the syndrome complain of vertigo, dizziness and loss of balance. The literature confirms that there are atypical forms of Usher syndrome type II characterized by, among others, vestibular dysfunction [1-3]. Other cases have also been reported in the scientific literature or anecdotally [7; 8]. Moreover, questionnaire-based studies show that individuals with retinitis pigmentosa (RP) may have vertigo and balance problems [4; 5]. Patients in the late stage of RP may present with visual destabilization of posture; this may stem from anomalous processing of visual information in the remaining visual field, as a result of the multiple retinal changes caused by RP [6].

IS THERE A LINK BETWEEN USHER SYNDROME TYPE II AND BALANCE PROBLEMS OR VERTIGO?

Usher syndrome is a genetic disease characterized by congenital deafness and progressive blindness caused by retinitis pigmentosa (RP). There are three forms. Type I is the most severe; it is manifested in profound congenital deafness, vestibular problems and childhood-onset RP. Type II is the most common form. It is characterized by moderate to severe congenital deafness, normal vestibular function, and RP typically appearing in the patient's late twenties or early thirties. Type III comprises progressive hearing loss, variable vestibular problems and RP [3].

As already mentioned, Type II Usher syndrome does not lead to vestibular problems. However, clinicians in the deaf-blindness program operated jointly by the Institut Nazareth et Louis-Braille and the Institut Raymond-Dewar have found that some patients with Type II Usher syndrome complain of vertigo, dizziness or loss of balance. According to the literature, there are atypical forms (genetic variations) of Type II Usher syndrome characterized, among others, by vestibular dysfunction [1-3]. Although these cases are rare, some have been identified in Tunisia, the United States and China. A Japanese study also reports a case presenting episodes of vertigo [8]. Anecdotal evidence is also to be found on on-line visual impairment forums, where some individuals with Type II Usher syndrome have reported dizziness or balance problems [7].

Apart from the fact that certain genetic variations of Type II Usher syndrome may lead to vestibular problems or vertigo, readers may want to know what role retinitis pigmentosa plays in these symptoms. In a study by Lowe & Drasdo (1992), many of the 48 subjects with RP reported experiencing vertigo in a variety of situations when walking, for example, when changing direction (n=9), avoiding other people (n=2), proceeding down a slope (n=1) or proceeding rapidly round the corner of a store window (n=1). In all, 20 of the 48 subjects reported symptoms of vertigo or confusion triggered by changing direction on a street or in a large public building [4]. Another questionnaire-based study was conducted in 2002 in Japan, among 828 persons with RP. The prevalence of cochlear symptoms and balance problems was 40.5%. Researchers speculate that balance problems in these individuals may be due to dysfunction of the vestibular, visual and central nervous systems [5]. Studies are needed to expand our knowledge of this topic.

In their literature review, Turano, Herdman & Dagnelie (1993) reported that postural stability is maintained by a combination of vestibular, somatosensory and visual cues. Visual cues include flow movement of the overall environment, modifications in image size, and retinal disparity. Motion information contributes majorly to visual stabilization

of posture, because it enables the observer to detect body oscillations relative to a stable background [6].

Turano, Herdman & Dagnelie (1993) conducted a study of 35 individuals with retinitis pigmentosa and 20 with normal vision but wearing eyeglasses simulating a visual field restricted to 26.5°, 15.5° and 6°. Subjects were asked to stand still while viewing a stable target or in darkness. The results showed that people with normal vision had better visual stabilization than those with RP. The authors also explain that as RP progresses, the visual stabilization index falls; values are normal when the disease first appears, but there is a linear reduction in visual stabilization as visual field restriction increases. However, they specify that loss of visual field alone does not wholly explain the findings of their study. One of the reasons for this statement is that in subjects with normal vision, artificial restriction of the visual field, even to 6°, did not cause postural destabilization. Turano et al. put forward the hypothesis that *in subjects with retinitis pigmentosa, visual destabilization of posture is the result of anomalous processing of visual information in the remaining visual field.*

Photoreceptors in the RP patient's retina deteriorate gradually, causing changes in spatial and temporal contrast sensitivity, acuity, motion detection thresholds and/or perception of spatial position. A combination of any of these changes may cause postural destabilization in these patients [6].

Conclusion

Based on the current medical classification, individuals with Type II Usher syndrome do not normally suffer from vestibular problems. Nevertheless, certain genetic variations may lead to an atypical form of the syndrome, characterized by vestibular dysfunction. It is also possible that retinitis pigmentosa contributes to balance problems, not only because of the reduction in visual field but also because of anomalous processing of visual information in the remaining visual field.

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