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EQUILIBRE ET VERTIGES

Fabry disease presenting cochleovestibular symptoms : case report with revue of the literature

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Abstract

Fabry disease (FD) is an X-linked lysosomal storage disorder caused by a deficiency in alpha-galactosidase A, leading to the accumulation of globotriaosylceramide (Gb3) in plasma and lysosomes. This aberration leads to a gamut of clinical complications, ranging from stroke/TIA to cardiac manifestations and renal failure. The otologic implications of Fabry disease (FD) have often been overlooked, despite their potential impact on quality of life. While not directly life-threatening, hearing loss and vertigo can significantly affect individuals with FD. A specific enzyme treatment has been developed over the last ten years, slowing the progression of the disease.

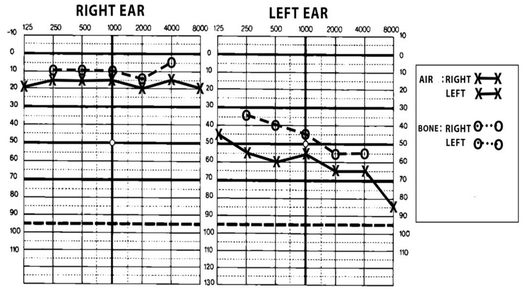
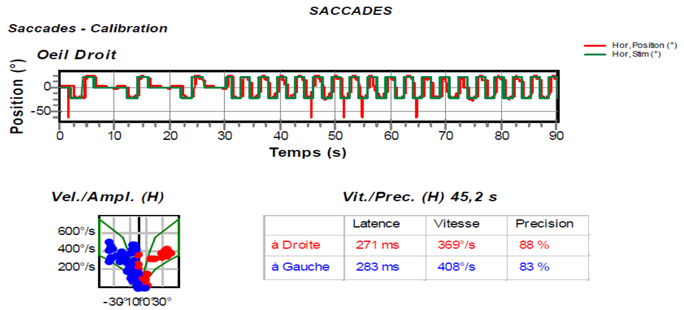


Figure1: Results of Pure tone audiometry



	Latence	Vitesse	Precision
à Droite	271 ms	369°/s	88 %
à Gauche	283 ms	408°/s	83 %

Figure 2: Results of saccades test

Objectifs

In this study we report a case of Fabry disease presenting cochleovestibular manifestations, with a review of the literature.

Méthodes et Matériels

In this study we report a case of Fabry disease presenting cochleovestibular manifestations

Résultats

Lorem Patient A.M., 22 years old, with a medical history of end-stage renal failure and chronic acroparesthesia, presented to our department with a 3-month history of left-sided hearing associated with right-sided tinnitus and vertiginous sensations, the whole evolving in a context of apyrexia and preservation of general condition. On examination, the patient was hemodynamically and respiratory stable. Neurological examination revealed hypersensitivity to thermal and pain stimulation. Otoscopy examination was normal. Vestibular examination revealed rotatory nystagmus, with a negative Fukuda test and head shaking test. The Dix and Hallpike maneuver was unremarkable. Saccadic movements were absent. Electromyography study was normal. The tympanogram and acoustic otoemissions were normal. Pure tone audiometry revealed a mixed hearing loss in the left ear, with an air-bone gap of 15 dB on 3 successive audiograms spaced of 15 days (Figure1). A Videonystagmography was performed and revealed left areflexia. Saccades were absent. (Figure 2) VHIT showed overt saccades. Based on the clinical presentation and the alpha-galactosidase enzyme assay revealing levels below 1%, the diagnosis of Fabry disease was confirmed. The patient benefited of an enzyme replacement therapy with the fitting of a conventional left-ear hearing aid. The patient also underwent 20 sessions of vestibular rehabilitation with good clinical evolution.

Conclusion

Fabry disease is a rare genetic disorder that warrants attention from otolaryngologists. Approximately 80% of patients experience cochleovestibular involvement, presenting as sensorineural hearing loss, sudden deafness, vertigo, or tinnitus. Regular auditory screening is essential for early detection of hearing impairment in Fabry patients. Early diagnosis is crucial for initiating enzyme replacement therapy promptly, which can mitigate irreversible organ damage and improve long-term prognosis. Otolaryngologists may be the first to suspect Fabry disease in cases of unilateral or bilateral sensorineural hearing loss in children, isolated tinnitus, or recurrent episodes of sudden deafness.

Références

1. Desnick RJ, Ioannou Y a., Eng CM. α-Galactosidase A Deficiency: Fabry Disease. OMMBID: The Online Metabolic and Molecular Bases of Inherited Diseases. 8th ed. 2013. p. α-Galactosidase A Defi- ciency: Fabry Disease. https://doi.org/10.1036/ommbid.181
2. Keshav S. Gastrointestinal manifestations of Fabry disease. NCBI Bookshelf. Oxford; 2006.
Macdermot KD, Holmes A, Miners AH. Anderson-Fabry disease: clinical manifestations and impact of disease in a cohort of 98 hemizygous males. J Med Genet. 2001; 38: 750–760. https://doi.org/10.1136/ jmg.38.11.750 PMID: 11694547