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Stikler's syndrome: A rare combined deafness and visual impairment

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Abstract

Aim: Stickler syndrome is a rare genetic disease of connective tissue that is among the main autosomal dominant syndromic deafnesses and constantly associated with a visual deficit. It also includes a rich ENT and general clinical features. The aim of our work is to report a rare case of this syndrome by describing deafness and various ENT manifestations that accompany it through an observation.

Materials and methods: We report the case of a little girl with Stickler syndrome and describe all ENT manifestations encountered, various additional examinations and therapy performed for her.

Results: 3-year-old girl, congenital cardiopathy, chronic bronchopneumonia, congenital hip dislocation and surgery for cleft lip and palate. She has myopia, right microphthalmia and chorioretinal coloboma. ENT signs: poor speech and recurrent rhinitis, particular facies with a flattened appearance, low-set ears and bilateral secretory otitis media. The tympanogram is flat. The stapedial reflex is absent on the left. ABR /ASSR asymmetrical deafness: profound with hearing reserves on low frequencies on the left and moderate transmissional profile on the right (50 dB). Insertion of a T tube bilaterally was made firstly. ABR/ASSR performed one month later: underlying moderate perceptive deafness on the right. A bilateral hearing aid with speech therapy was recommended, providing satisfactory hearing gain as well as a clear improvement in the quality of language.

Conclusion: Stickler syndrome should be known by otolaryngologists. Constant facial dysmorphism, oropharyngeal anomalies and deafness allow to evoke its diagnosis and to establish early management. Its suspicion should lead to an ophthalmological examination in order to correct associated visual impairment.

Keywords

“Stickler syndrome; deafness; myopia; ABR/ASSR; cleft palate”.

Biography

-MEHTARI NASRINE SIHAM, born on December 13, 1981 in Oran Algeria

- ENT doctor since 2010

- Assistant professor at the Tlemcen University Hospital since 2011

- University hospital professor since 2023

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