HEARING IMPAIRMENT IN CHILDREN WITH STICKLER SYNDROME: REVIEW OF PHENOTYPE AND CORRELATION WITH GENOTYPE

Acke F, Dhooge I, De Leenheer E
Ghent University / Ghent University Hospital, Department of Otorhinolaryngology
Research Foundation Flanders

OBJECTIVES: Stickler syndrome is a connective tissue disorder characterized by ocular, skeletal, orofacial and auditory defects. Autosomal dominant Stickler syndrome is caused by mutations in different collagen genes, namely COL2A1, COL11A1 and COL11A2. In light of our own future study concerning hearing impairment and the link with mutated gene and mutation type in Stickler patients, a review of the literature on the subject is presented. METHODS: English-language literature was reviewed through searches of PubMed and Web of Science, in order to find relevant articles describing auditory features in Stickler children aged 0-17, along with genotype. RESULTS: 103 patients (68 families) described in 34 articles were included. Hearing loss was found in 69 patients (67.0%), mostly mild to moderate when reported. Hearing impairment was predominantly sensorineural (52.5%), followed by mixed (27.1%) and conductive (20.3%). All patients with conductive and most with mixed hearing loss (78.6%) had a palatal defect, while this was 42.3% in children with sensorineural hearing impairment. Defects in COL11A1 (81.3%) en COL11A2 (100.0%) are more frequently associated with hearing loss than mutations in COL2A1 (56.9%). CONCLUSIONS: Hearing impairment in children with Stickler syndrome is common. As in adults, sensorineural losses predominate. However, in children, a conductive component is also frequently observed (47% vs. 22% estimated in adults), especially in patients with a palatal defect, prone to develop chronic otitis media. Strict follow-up of young Stickler patients is strongly advised. The distinct disease-causing collagen genes lead to a different risk of hearing impairment, but still large phenotypic variation exists.