

# HEARING IMPAIRMENT IN STICKLER SYNDROME: A SYSTEMATIC REVIEW

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## Abstract

**OBJECTIVES:** Stickler syndrome is a connective tissue disorder characterised 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 the mutated gene and mutation type in Stickler patients, a systematic review of the literature on the subject is presented. **METHODOLOGY:** English-language literature was reviewed through searches of PubMed and Web of Science, in order to find relevant articles describing auditory features in Stickler patients, along with genotype. The systematic review was conducted according to the PRISMA guidelines. **RESULTS:** 315 patients (108 families) individually described in 40 articles were included. Hearing loss was found in 63.8%, mostly mild to moderate when reported. Hearing impairment was predominantly sensorineural (70.2%). Conductive (12.2%) and mixed (17.7%) hearing loss was primarily found in patients with young age or a history of palatal defect. Overall, mutations in *COL11A1* (80.0%) en *COL11A2* (93.3%) seem to be more frequently associated with hearing impairment than mutations in *COL2A1* (54.2%). **CONCLUSIONS:** Hearing impairment in patients with Stickler syndrome is common. Sensorineural hearing loss predominates, but also conductive hearing loss, especially in children and patients with a palatal defect, can occur. The distinct disease-causing collagen genes lead to a different prevalence of hearing impairment, but still large phenotypic variation exists. Regular auditory follow-up of Stickler patients is strongly advised.