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Hearing loss and connective tissue disorders in Stickler syndrome

Célia Alcobia Gomes

Instituto Politécnico de Coimbra, ESTeSC-Coimbra Health School, Portugal

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Background: Stickler syndrome is a group of hereditary conditions in the collagen connective tissue that can include a distinctive facial appearance, hearing loss, eye abnormalities, and joint problems. A characteristic feature of Stickler syndrome is a somewhat flattened facial appearance. The hearing loss is both conductive and sensorineural and varies in degree and may become more severe as age increases. Stickler syndrome affects an estimated 1 in 7,500 to 9,000 newborns. The diagnosis of Stickler syndrome is clinically based, but understanding the genetic basis of this syndrome is important because numerous independent genes have been identified. Stickler syndrome can be inherited in an autosomal dominant manner or in an autosomal recessive manner depending on the gene that has a change (mutation or pathogenic variant). When Stickler syndrome is caused by pathogenic variants in *COL2A1*, *COL11A1*, or *COL11A2* genes, it is inherited in an autosomal dominant manner. Pathogenic variants in *COL9A1*, *COL9A2*, or *COL9A3* genes cause Stickler syndrome inherited in autosomal recessive manner. Between 80-90% of all cases are classified as type I and are caused by mutations in the *COL2A1* gene. Another 10-20% of cases are classified as type II and result from mutations in the *COL11A1* gene.

Material and methods: Using molecular genetic techniques, the identification of genes associated with hearing impairment in Stickler syndrome will allow screening and possible therapy for the hearing impaired. An early diagnosis can be made and early intervention can be pursued to establish, preserve, or restore functional hearing to maximize speech-language development in the setting of verbal communication, and allows genetic counseling for the families.

Results: The typical structure of collagen, a triple helix of three procollagen fibrils allows it to perform the cell adhesion function, strengthening the structures where it is present. If one of the fibrils is missing, the protein complex cannot fulfil its function in the living cell, and the cell probably have an abnormal function and the symptoms of Usher\' syndrome appear.

Conclusions: Describe the presentation and genetics of the Stickler syndrome, one of the most frequently occurring syndromes involving hearing loss and help the correct diagnosis and treatment.

Conflicts of interest: None.