



ISIC-2022 International Scientific Interdisciplinary Conference





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reduction of HDL-C was observed in women. But more significant elevation of CRP was observed in men, which indicates higher level of systemic inflammation.

Conclusions: Changes of lipid profile in patients with diabetes mellitus 2 type obviously depend on gender. The mean levels of TC, LDL-C and TG appeared to be statistically significantly higher, while HDL-C levels were significantly lower in women in comparison with men. Further investigations in order to reveal the etiology of gender differences in lipid profiles in patients with DM-2 and new approaches to the treatment of dyslipidemia according to the gender peculiarities are needed.

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STICKLER SYNDROME

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Introduction. The similar phenotypic features of certain inherited connective tissue disorders (e.g., Marfan, Ehlers-Danlos, and Beals syndromes) makes it difficult to differentiate them and make a true diagnosis. One of these diseases is Stickler syndrome.

Aim. Determine the most common symptoms and the importance of the early diagnostics of Stickler syndrome.

Material and methods. We conducted a literature review and comprehensive analysis of the following databases: EMBASE, PubMed, The Cochrane, NHS Centre for Reviews and Dissemination.

Conclusions. Stickler syndrome belongs to systemic hereditary connective tissue diseases, specifically to collagenopathies and accompanied by a disruption in the structure and function of collagen types 2, 9 or 11. According to the modern classification, there are 5 types of Stickler syndrome – 5 types of mutations in the genes responsible for collagen synthesis: COL2A1, COL11A1, COL11A2, COL9A1, COL9A2. Due to the similarity of their symptoms and common therapeutic approaches, we will consider only the first type of this syndrome, the most common



one. Defective collagen is expressed as a result of COL2A1 gene damage, which affects the relevant tissues' structure and function, e.g. bone, cartilage, tendon tissue, myocardium or vitreous. The clinical symptoms are musculoskeletal system and ocular disorders, which reflects an alternative name for the syndrome - congenital progressive artroophthalmopathy. The main manifestations are progressive myopia, which manifests in the first decade of life and ends with retinal detachment, as well as premature degenerative changes in various joints and expansion of the epiphyses according to radiography. Other ocular disorders include proliferative vitreoretinopathy, astigmatism, cataracts, strabismus and glaucoma. Changes in the musculoskeletal system are characterized by early onset of joint pain and stiffness, their primary hypermobility, followed by the gradual formation of joint degeneration. Stickler's syndrome patients have a flattening of the middle part of the face, which is caused by maldevelopment of this area of the facial skeleton, in particular zygomatic bones and nose bridge. Facial anomalies also include small size of the upper and lower jaws, a protruding upper lip, soft and hard palate cleft, often patients have a marfanoid phenotype. Hearing loss is one of the main features as well. Mostly it does not require the use of hearing aids. Approximately half of the patients have mitral valve prolapse. In general, children's psychoverbal development conforms to the norm; however, there are cases of mild to moderately decreased level of intelligence. Stickler's syndrome patients are primarily treated surgically, which include cryotherapy and laser treatment of retinal detachment, plastic surgery of the soft and hard palate and joint prosthetics. Early diagnosis helps prevent life-threatening complications such as retinal detachment, deafness and even heart failure.