

# ENT conditions and disorders in children with Hunter syndrome

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**Objective.** To describe the features of the ENT conditions and disorders in children with Hunter syndrome. Hunter syndrome, also known as mucopolysaccharidosis type II (MPS II), is a rare X-linked recessive disease. The MPS II is a hereditary metabolic disorder caused by accumulation of glycosaminoglycans (GAG) in organs and tissues due to mutations in the genes, which encode intralysosomal hydrolysis of macromolecules. Hunter syndrome is a progressive, multisystem disease. At the same time a child may have mental retardation and speech development delay, skeletal bone deformities, loss of vision, hearing loss.

**Methods.** The study included 17 boys aged from 2 to 12 years old with genetically confirmed Hunter syndrome. All patients underwent examination by otolaryngologist, also tympanometry, diagnostic nasopharyngolaryngoscopy, registration of otoacoustic emission, audiometry, cardiorespiratory monitoring were performed.

**Results.** The accumulation of GAG leads to a gradual narrowing of the nasopharynx and larynx lumen, a thickening of the tongue and vocal folds, laryngeal cartilages deformation, an enlarged of pharyngeal, palatine and lingual tonsils.

11 children (64.7%) had chronic inflammation of the nasal mucosa and pharyngeal tonsil with frequent exacerbations. Pharyngeal tonsil hypertrophy was detected in 10 patients (58.8%).

14 children (82%) had a wide, thickened tongue. Hypertrophy of the tonsils was diagnosed in 8 children (47%).

The voice had changed (hoarseness) in 16 children (94%). Deformation of the epiglottis was detected in 4 people (23.5%), tracheomalacia — in 1 child (5.8%).

Among the complications acute sinusitis and exudative otitis media were detectable in 52.9% of patients. 10 children (58.8%) had obstructive sleep apnea (OSA) syndrome, two of them (11.7%) had a severe degree of the disease, which was an indication for adenotonsillectomy. Hearing loss was found in 10 boys (58.8%).

In most patients (82%) a combined pathology of the ear, throat and nose was detected, 2 children (11.7%) had only adenoid hypertrophy, and only 1 child (5.8%) did not have any pathology of ENT organs.

**Conclusion.** Functional disorders and diseases of the ear, throat and nose are found in most children with Hunter syndrome, which could be one of its early manifestations. An ENT specialist may suspect MPS type II according to the presence of an ENT pathology that is difficult to treat in a standard manner, combined with the pathology of other organs and systems.

Early diagnosis and the possibility of enzyme replacement therapy can control the disease progression and avoid early disability.