Abstract

Objective: Otorhinolaryngological (ENT) symptoms are frequent and often the first signs of lysosomal storage diseases (LSD). The most common are otological problems, adenotonsillar hypertrophy and progressive airway obstruction. The aims of this work were to evaluate ENT symptoms and their occurrence in a group of Czech patients with mucopolysaccharidosis (MPS), to describe the risks and complications associated with administration of general anaesthesia, and to characterize adenotonsillar tissue changes in patients with MPS.

Methods: Patients with a genetically confirmed MPS were included in the studies. Within the entire group of LSDs, the work is focused on the subgroup of MPS.

Results: We published a study of 61 patients with different MPS types. We described ENT manifestations and surgical procedures in detail. We identified the time relationship between ENT presentation, first ENT procedure and the age at which the MPS diagnosis was established. At least one ENT symptom was present in 90 % and ENT surgery was performed in 57 % of patients. In three quarters of patients, ENT symptoms appeared before the diagnosis of MPS. The median age at the time of the first ENT symptom was in all MPS subtypes lower than the median age at the time of MPS diagnosis. The most common ENT manifestations were chronic/recurrent rhinosinusitis (77 %), upper airway obstruction (65 %) and hearing impairment (53 %). Adenotomy was the most common surgical procedure (56 %).

In MPS II patient, we described the insertion of a tracheal stent to resolve tracheomalacia. We summarized recommendations for managing anaesthesia in MPS patients.

In a histopathological study, we characterized adenotonsillar tissue changes in MPS using histological, immunohistochemical, immunofluorescence and ultrastructural analyses. We identified a new (immuno)histochemical and ultrastructural phenotype of lysosomal storage changes in a specific subtype of adenotonsillar paracortical cells in 8 MPS patients. Abnormal cells were effectively detected using an antibody targeting the lysosomal membrane tetraspanin CD63. These CD63 positive storage vacuoles lacked the monocyte/macrophage marker CD68.

Conclusions: High and early occurrence of ENT symptoms in MPS patients highlights the role of otorhinolaryngologists in prompt recognition of the disease allowing early initiation of a specific therapy and effective follow-up of these patients. Histopathological examination of the lymphoepithelial tissue can, due to typical findings, facilitate diagnosis and help evaluate the effects of MPS therapy.