Ocena fenotypowa pacjentów z zespołem Pendreda

Phenotypic evaluation of patients with Pendred syndrome

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Introduction. The Pendred syndrome (PS) is an autosomally recessively inherited disease. Its diagnosis requires identifi cation of the classical triad of symptoms, including hypoacusis, thyroid goitre and iodine organifi cation defect in the thyroid, which may lead to thyroid functional disorders of hypothyroidism. SP is accompanied by anatomical anomalies. The objective is the hearing and balance system evaluation and the analysis of the inner ear structure and also the assessment of the function and structure of thyroid gland. Material and methods. For the research four families were qualifi ed, 7 persons with PS, 12 persons altogether. In all the patients the anamnesis in the form of a questionnaire and laryngological examination were performed. It was followed by pure tone, speech and impedance audiometry and brainstem response testing as well. ENG was also conducted. Patients with hearing loss were subjected to magnetic resonance of temporal bone. For the whole group thyroid hormones levels and iodine organifi cation in the thyroid identifi ed in a test with potassium perchlorate were measured and also USG and scyntography were conducted. Results. In audiological examination in 3 cases deafness, in 2 cases profound hypoacusis and in 2 mild hypoacusis were recognised. In the group in 2 patients the hypoacusis was of a mixed type. In radiological assessment the labirynth showed anatomical anomalies in the form of enlargement of the vestibular aqueduct and the endolyphatic sac, yet in 3 patients the anomalies also concerned the structure of cochlear and semicircular canals. Endocrine examination showed hypothyroidism in 5, its subclinical form in 1, diffuse thyroid goitre in 4 and nodular thyroid goiter in 2 cases. Conclusions. A complex clinical evaluation: endocrine and audiological, together with radiological diagnostic imaging, supported by molecular studies of SLC26A4 gene, are the procedures, necessary for complete and accurate diagnosis of PS and EVAS.