

Background. Multiple endocrine neoplastic syndromes (Multiple Endocrine Neoplasia - MEN) are a group of disorders characterized by simultaneous or successive association of hyperplastic or tumoral lesions, usually benign or malignant, usually hypersecretory, at least involving two endocrine glands, without obvious functional interrelations. MEN 1 is one of the three distinct types of MEN that affect the parathyroid glands, endocrine pancreas and anterior pituitary gland. Parathyroid glands are the first and most commonly affected by MEN 1 (95%). The diagnosis can be established before the age of 20 by identifying some suggestive signs – recurrent urinary lithiasis, muscle or bone pain. All patients with MEN should be registered and supervised throughout life, given that the tumors remain asymptomatic for a long time.

Case report. Patient X, age 48, has been suffering for approximately 20 years for kidney stones. Was operated on parathyroid, then had consecutive surgeries on the kidneys, and in 2015 started the treatment with hemodialysis. In the meantime, hyperprolactinemia has been identified, and pituitary adenoma has been described on MRI. For the moment, the Patient X is on a complex pathogenic treatment.

Conclusions. 1. Parathyroid adenoma is commonly found in polyglandular neoplastic syndromes. 2. Hyperparathyroidism once diagnosed, it is necessary to differentiate primary, secondary or tertiary type in order to establish prompt treatment tactics and to prevent complications. 3. The complex approach of the patient with hyperparathyroidism is essential.

Key words: hyperparathyroidism, MEN 1, pituitary adenoma, hemodialysis.

DEPARTMENT OF PEDIATRIC SURGERY, ORTHOPEDICS AND ANESTHESIOLOGY

4. ASSOCIATION OF ULTRASHORT SEGMENT HIRSCHSPRUNG DISEASE WITH A RARE GENETIC PATHOLOGY – TOWNES-BROCKS SYNDROME

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Background. In our case we have find an association of two different pathologies - Townes-Brocks syndrome (TBS) and Hirschsprung's disease (HD). TBS is a rare autosomal dominant disease (1/250000 newborns), caused by mutation of the gene SALL1. There is approximately 200 described cases in the special literature. The main symptoms are: malformations of the external ear (by dimensions, form, insertion, presense of preauricular rudimentary tags), anorectal malformations (imperforated anus, anal atresia, anal stenosis), heart and renal malformations, limb's abnormalities (three-articular thumb, syndactyly, overlapping fingers). TBS can be diagnosticated in base of simultaneously presense of major symptoms triad, sometime without genetic research. The Ultrashort Segment HD is a controversial form of aganglionosis wich involves 2-4 cm of distal rectum and anal canal. It is characterized by latent debut, lack of classical radiological interpretation and negative recto-anal reflex by manometry. The treatment of Ultrashort Segment HD is also controversial. Some

authors prefer posterior anal sphincter myectomy, and some prefer excision of the aganglionic segment and pull-through reconstruction.

Case report. Here, we present the case of 1-year-old Moldavian boy born with anal atresia (with fistula in situ), low placed and small external ears, three-articular thumbs and overlapping fingers of the legs. Anoplasty was performed at 2-months-old without complications. Further was installed persistent constipation. Imagistic, functional and histological investigations revealed Ultrashort-Segment HD with left megadolichocolon. We performed abdominoperineal resection of left colon by Swenson-Pellerin with coloanal anastomosis. After 6 months postoperatively symptoms of colonic retention or dyssinergic defecation are absent. A karyotype research demonstrated only a normal male chromosome constitution (46XY) with a dense site ADN gh(+)_{18q}, but molecular gene-testing actually cannot be executed in Moldova, and the TBS was established phenotypically in base of major symptoms triad.

Conclusions. Association of Ultrashort Segment HD with TBS doesn't influence the surgical tactic of HD, but represents an interest in treatment, functional and social rehabilitation and staged correction of concomitant malformations.

Key words: Hirschsprung disease, Townes-Brocks syndrome

5. RADIOLOGICAL AND CLINICAL CONFLICTS IN A CASE OF TRANSPOSITION OF THE GREAT ARTERIES WITH MULTIPLE ASSOCIATED COMORBIDITIES

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Background. Transposition of the great arteries is an embryological misplacement of the Aorta and the trunk of the Pulmonary Artery, in which the Aorta rises from the right ventricle, while the pulmonary trunk continues the left ventricle, thus creating two parallel vascular systems. This situation is not compatible with life in the absence of a communication between the two systems (e.g. Ventricular septum and/or atrial septum defect, persistence of the arterial duct etc.) which will allow the mixing of oxygen-rich blood with deoxygenated blood. In order for the patients to survive, this congenital heart disease has to be treated as soon as possible. In some circumstances the surgery can be post-poned by using prostaglandines to keep the arterial duct open. Considering this information, we decided to look upon a case of TGA with multiple comorbidities and evaluate the role of radiologic and ultrasound(US) investigations in decisions regarding the tempos of the multidisciplinary surgical interventions.

Case report. We will present the case of a newborn female, prenatally diagnosed with TGA, who was transferred from another clinic, where an ileostomy was performed, to temporarily treat her inability to feed. She associated a diaphragmal hernia, metabolic uncompensated acidosis, anemia, elevated respiratory rate, fever and decrease of SpO₂. She was treated with PGE₁ prior to the surgical interventions which took place in our clinic. After her admission paraclinics confirmed the TGA and diaphragmal hernia through repeated radiographies, and identified the need of closing the ileostomy and reconstructing the digestive tract, due to the presence of peritonitis seen during ultrasound investigations. The patient has undergone a complex multidisciplinary surgical intervention, with the aim of simultaneously fixing all the cardiac and digestive abnormalities through toracotomy aswell as laparotomy. The decision of