A 10-year-old girl with primary hypoparathyroidism and systemic lupus erythematosus (SLE)

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Introduction:

Parathyroid Hormone (PTH) is one of the principal regulatory hormones for calcium and phosphate homeostasis. Reduced PTH concentration in hypoparathyroidism is characterised by persistent hypocalcaemia and hyperphosphataemia. Hypoparathyroidism in children can occur either as a component of a genetic syndrome e.g. 22q Deletion (DiGeorge) Syndrome, autoimmune disorder or be acquired secondarily to thyroidectomy or some destructive processes of the glands.

Case presentation:

We report a ten-year-old girl who initially presented to the Department of Pediatric Neurology and Rehabilitation, Medical University of Białystok due to repeated seizures, but then, after hypocalcaemia had been confirmed, was admitted to the Department of Paediatrics, Endocrinology, Diabetology with Cardiology with suspected hypoparathyroidism. Her postnatal medical history was unrevealing, and there was no history of candidiasis. Regarding family history, patient’s mother reported epilepsy and arrhythmias.

The girl was admitted severely unwell with drowsiness and confusion. Physical examination revealed a non-specific rash on the whole body surface (probably an allergic reaction to oxcarbazepine), and mild insignificant dysmorphic features - hypotelorism. Because of the low PTH concentration (<3pg/ml; NR 10-60) and typical biochemistry (total serum calcium concentration 0.8 mmol/l, plasma phosphate 4.1 mmol/l), primary hypoparathyroidism was confirmed. Other hormonal analyses showed no thyroid or adrenal disorders (TSH, FT4, cortisol and ACTH level were normal). Liver and kidney function were normal. Ultrasonography of the thyroid and parathyroids showed a hyperechoic area in the thyroid left lobe (II criteria). The patient was initially treated with intravenous and oral calcium, vitamin D3 and alfalcacidol. Sevelamer, a phosphate-binding drug was used for the management of hyperphosphataemia. The patient was also treated with valproic acid for the concomitant epilepsy and beta-blocker for LQTc syndrome. Calcium in the serum remained below the normal reference range, and phosphate level increased initially despite the treatment.

During hospitalization the girl twice developed intermittent fever, accompanied by an elevated CRP and radiological features of pneumonia and pericardial effusion. Despite negative blood cultures and no serological evidence of viral infections, including HIV, B19 parvovirus, influenza virus, a number of zoonoses and tuberculosis she was treated empirically with antibiotics.

The CT scan, MRI and PET MRI did not reveal any abnormalities. Bone mineral density measured with DXA was within the normal range. Based on the presence of standard clinical criteria, systemic lupus erythematosus (SLE) was diagnosed and the treatment with glucocorticoids was initiated, which improved parameters of calcium-phosphate balance.

Further immunological examinations revealed INF-omega antibodies before implementation of steroid therapy, with negative IL-22, IL-17A, IL-17F, IFN-lambda, IFN-omega, IFN-alpha2A, and CaSR antibodies after this treatment. Genetic diagnosis excluded AIRE and CaSR mutations.

Coexistence of hypoparathyroidism with SLE, the presence of INF-omega autoantibodies and normalization of calcium and phosphate serum concentration following glucocorticoid treatment may suggest an autoimmune background of the disease in the patient. However, further investigation is needed to detect specific underlying mechanism responsible for insufficient PTH secretion.

Conclusions:

Autoimmune antibodies screened for in the patient