

Idiopathic hypoparathyroidism in a 10-year-old girl with concomitant epilepsy, Long Q-T Syndrome (LQTS), pericarditis and pneumonia.

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Introduction:

PTH is one of the principal regulatory hormones for calcium and phosphate homeostasis. Hypoparathyroidism, caused by reduced parathyroid hormone (PTH) concentration is characterised by hypocalcaemia and hyperphosphataemia. Hypoparathyroidism in children can occur either as part of a genetic syndrome, autoimmune disorder, be acquired secondarily to thyroidectomy or some destructive process of the glands.

Case presentation:

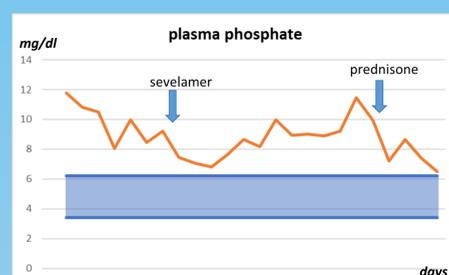
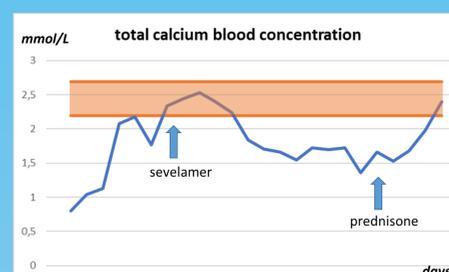
We present a ten-year-old girl who initially presented to the Department of Paediatric Neurology and Rehabilitation, Medical University of Białystok with convulsions but then, after hypocalcaemia was confirmed, admitted to the Department of Paediatrics, Endocrinology, Diabetology with Cardiology with suspected hypoparathyroidism. There was no history of candidiasis. In the family history, mother had epilepsy and arrhythmias. She was admitted severely unwell with drowsiness and confusion. The physical examination revealed a rash on the whole body (probably an allergic reaction to oxcarbazepine), caries, mild dysmorphic features -hyperthelorism. Because of the low PTH concentration (<3 pg/ml) and typical biochemistry (total calcium blood concentration 0.8 mmol/l, plasma phosphate 4.1 mmol/L) hypoparathyroidism was confirmed. Other hormonal analyses showed no thyroid or adrenal gland disorders (TSH, fT4, cortisol, ACTH level normal). Liver and kidney function were normal. Ultrasonography of the thyroid and parathyroids showed a hyperechoic area in the thyroid left lobe, but no parathyroid pathology. Ultrasonography of abdomen and echocardiography were normal. Long QTc (over 0.5 seconds) was present in the ECG.

Hypocalcemia was initially treated with intravenous and oral calcium, vitamin D₃ and the synthetic precursor of the active form of vitamin D₃, alfacalcidol. Hyperphosphataemia was treated with sevelamer, a phosphate-binding drug. She was also treated with valproic acid for epilepsy and hydrocortisone to reduce her allergy symptoms. Calcium remained low and phosphate raised initially despite treatment until sevelamer was added after which calcium and phosphate started to normalise. During hospitalisation she developed intermittent fever, raised CRP and radiological features of pneumonia and pericardial effusion. Despite negative blood cultures and no evidence of viral infections, including HIV, B19 parvovirus, influenza virus, many zoonoses and tuberculosis she was treated empirically with antibiotics. PET MRI didn't reveal any pathology.



pericardial effusion

Further diagnosis revealed INF- ω autoantibodies but no 21-hydroxylase autoantibodies. Rheumatological consultation suggested lupus erythematosus and the treatment with glucocorticoids was started that improved calcium and phosphate metabolism.



Conclusions:

Coexistence of hypoparathyroidism with pericarditis and pneumonia is unusual. Sevelamer proved useful in correcting the plasma phosphate more rapidly than alfacalcidol alone but glucocorticoid treatment of coexisting rheumatological disease have the best positive effect on calcium and phosphate metabolism. A diagnosis of idiopathic hypoparathyroidism may sometimes be modified after other typical symptoms following initial presentation. Our patient will be observed in the direction of autoimmune polyglandular syndrome.

ACKNOWLEDGEMENTS

Jadwiga Furmaniak and Bernard Rees Smith, FIRS Laboratories, Cardiff, UK for assasing INF- ω and 21-hydroxylase autoantibodies

Piotr Gietka and Elżbieta Dobreńko for rheumatologic consultation