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Hypoparathyroidism with Cartwheel-like Change in Thoracic Vertebral Body

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Abstract A 34-year-old man was transported by ambulance to our hospital after experiencing generalized convulsions. He had been of short stature since birth, and had been diagnosed with growth hormone deficiency, which was treated with growth hormone therapy from 10 to 14 years of age. He experienced convulsions two times at 32 and 33 years of age, respectively, with spontaneous recovery. He had no specific family history. On arrival, he had clear consciousness and sinus tachycardia. A physical examination revealed short stature. A biochemical analysis of venous blood revealed hypocalcemia and a low level of parathyroid hormone. Head computed tomography (CT) revealed diffuse hyperostosis of the cranial vault and truncal CT revealed cartwheel apperance in the thoracic vertebral bodies from Th9 to Th12. He was treated with levetiracetam, alfacalcidol and calcium agents. Next-generation sequencing suggested heterozygous large deletion of T-box transcription factor (TBX1) gene. This is the first report of hypoparathyroidism with cartwheel-like change in the thoracic vertebral body. The further accumulation of cases is necessary to determine whether this change is specific to hypoparathyroidism.

Keywords: *convulsion, hypocalcemia, skull hyperostosis, computed tomography*

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1. Introduction

Hypoparathyroidism is a rare disorder of parathyroid hormone (PTH) deficiency, which is regulated by the calcium-sensing receptor (CaSR) found on parathyroid chief cells, with an estimated prevalence of 23-37 cases per 100,000 person-years. [1] It most commonly occurs after anterior neck surgery; however, there is an expanding list of genetic and nonsurgical acquired causes. [1] PTH is critical for maintaining the level of circulating calcium through its actions in bone, as well as in the kidneys and intestines. [1,2,3] Most patients with hypoparathyroidism have neuromuscular signs and symptoms of hypocalcemia, which include mild paresthesia, muscle cramps, prolongation of the corrected QT interval, arrhythmias, laryngospasm, and seizures. [1,2,3] Many patients have a reduced quality of life, with nonspecific symptoms that can include general fatigue, a lack of focus, depression, and other neuropsychiatric issues. [1,2,3] Basal ganglia calcification, cataracts, fracture or renal complications are a feature of longstanding nonsurgical hypoparathyroidism. [1]

We herein report a case of nonsurgical hypoparathyroidism after convulsion, in which the patient showed cartwheel-like change in thoracic vertebral body and skull hyperostosis.

2. Case Presentation

A 34-year-old man was transported by ambulance to our hospital after experiencing generalized convulsions while playing pachinko. The patient had short stature since birth. He was diagnosed as growth hormone deficiency, and was treated with growth hormone therapy from 10-14 years of age at a pediatric hospital. Based on his mother's statement, his scholastic attainment had been poor, he also had a short temper, difficulty in human relationships, and never had a fixed job. He experienced convulsions two times at 32 and 33 years of age with spontaneous recovery, and did visit a medical facility at that time. He had no specific family history. On arrival, his consciousness was clear and his vital signs were as follows: blood pressure, 134/86 mmHg; heart rate, 108 beats per minute; respiratory rate, 20 breaths per minute; percutaneous saturation, 98% under room air; and bladder temperature, 36.3°C. On physical examination, he had short stature (158 cm, 56 kg). A biochemical analysis of venous blood on arrival revealed the following: pH, 7.344; PCO2, 35.0 mmHg; PO2, 73.1 mmHg; HCO3-, 18.6 mmol/L; base excess, -5.9 mmol/L; lactate, 6.6 mmol/L; white blood cells, 5,100/μL; hemoglobin (Hb), 14.5 g/dL; platelets, 20.5×10^4/μL; total protein, 7.8 g/dL; albumin, 4.4 g/dL, glucose, 136 mg/dL; HbA1C, 5.7%; total

bilirubin, 0.6 mg/dL; aspartate aminotransferase, 25 IU/L; alanine aminotransferase, 17 IU/L; alkaline phosphatase, 242 IU/L; γ-glutamyltranspeptidase, 11 IU/L; blood urea nitrogen, 10.3 mg/dl; creatinine, 0.89 mg/dl; creatine phosphokinase, 509 IU/L; uric acid, 11.8 mg/dL: sodium, 142 mEq/L; potassium, 3.6 mEq/L; calcium, 6.5 mg/dL; phosphate, 2.4 mg/dL; prothrombin time 11.4 (11.4) sec; activated partial thromboplastin time, 24.0 (26.9) sec; fibrinogen, 243 mg/dL; fibrin degradation product 2.4 μg/ml and CRP, 0.15 mg/dl. The results of a urinalysis were within the normal limits. Head computed tomography (CT) revealed diffuse hyperostosis of the cranial vault with calcification at the globus pallidus and choroid plexus (Figure 1).

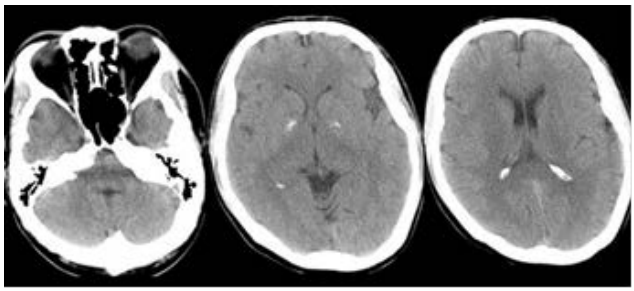

Figure 1. Head computed tomography (CT) (Head CT shows hyperostosis of the cranial vault with calcification at the globus pallidus and choroid plexus)

The width of the frontal bone was 9.7 mm (normal range 3.5 ± 0.3 mm).[4] Truncal CT revealed a cartwheel appearance in the thoracic vertebral bodies from Th9 to Th12 (Figure 2).

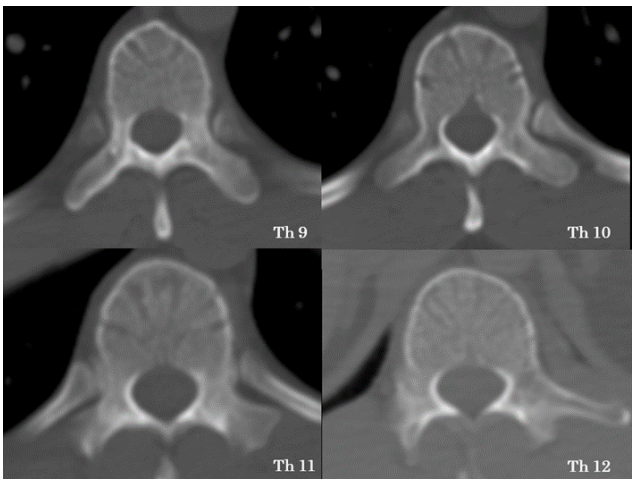

Figure 2. Thoracic vertebral body (The thoracic vertebral bodies from Th9 to Th12 show a cartwheel apperance)

As he was tentatively diagnosed with convulsion due to hypocalcemia, he was admitted and treated with levetiracetam, alfacalcidol and calcium agents. He temporally showed muscle pain due to cramp; however, this subsided after the improvement of hypocalcemia. Later, the following findings were observed: rheumatoid factor, 1.7 (0-15) IU/mL; anti-nuclear antibody < 40; PTH-intact, 7 (10-65) pg/mL; vitamin D, 16.6 (10~70) ng/mL; 1,25-dihydroxyvitamin D3, 46 (20-60) pg/mL, growth hormone, 1.01 (0-2.47) ng/mL; insulin-like growth factor-1, 98 (102-283) ng/mL. Electroencephalography and brain magnetic resonance imaging did not reveal any specific findings. A WAIS-III test revealed the following of verbal intelligence quotient (IQ), 25; performance IQ, 12; and full scale IQ, 37 (verbal comprehension, 13; perceptual organization, 7; working memory, 16; and processing speed, 6). After his symptoms subsided, he was discharged and followed as an outpatient. Next-generation sequencing (target sequencing using hybrid capture methods) suggested heterozygous large deletion of the T-box transcription factor (TBX1) gene, which is the major candidate gene for 22q11.2 deletion syndrome, which can cause hypoparathyroidism.

3. Discussion

This is the first report of hypoparathyroidism with cartwheel-like change in the thoracic vertebral body. The hypoparathyroidism in the present case is thought be genetic; thus, hypocalcemia might have been present since birth. In addition, his short stature might have been induced by TBX1 gene abnormality, rather than growth hormone deficiency. However, he was diagnosed with hypoparathyroidism at 34 years of age after experiencing convulsions. Nonsurgical hypoparathyroidism is rare and is frequently represented by neuropsychiatric disorders; thus, the misdiagnosis rate of the disease is high.[5] Accordingly, physicians should evaluate calcium levels when treating convulsive patients. Given that even genetic hypoparathyroidism can be associated with a late onset, the present case might represent a case of late onset hypoparathyroidism. [6]

The vertebral venous channels constitute a network of conduits that traverse the cancellous and cortical bone of normal vertebral bodies. These structures are readily demonstrated on high-resolution computed tomography (Figure 3). [7] The normal anatomy of the basivertebral veins in the axial plane shows a V-shaped configuration of major basivertebral venous channels in the thoracic vertebral body and a Y-shaped configuration with a prominent central confluence and anterolateral cortical perforations in the lumbar vertebral body. [8,9]

To the best of our knowledge, the multiple radiating pattern of the anterior channels with cortical fenestrations (cartwheel-like change) has not been reported. As hypoparathyroidism is associated with increased bone density and the macroscopic and microscopic skeletal features reflected a low turnover state, these features could produce cartwheel-like changes in the vertebral body. [10,11] The further accumulation of cases is necessary to determine whether this change is specific to hypoparathyroidism.

Regarding hyperostosis of the skull, TBX 1(-/-) mice display short stature, the absence of a hyoid bone, failed fontanelle closure, a bifid xiphoid process, and hypoplasia of the clavicle and zygomatic arch. [12,13] The loss of TBX 1 in mice (TBX1 (-/-)) results in skeletal abnormalities similar to those of cleidocranial dysplasia (CCD) in humans. [12] However, we could not find any TBX1 gene abnormality or hypoparathyroidism could manifest hyperostosis of cranium. Patients with hyperparathyroidism or excess of growth hormone can show hyperostosis of the cranium. [14,15] As the present case underwent growth hormone treatment at 10-14 years of age, this might have resulted in the onset of hyperostosis of the cranium in the present case.

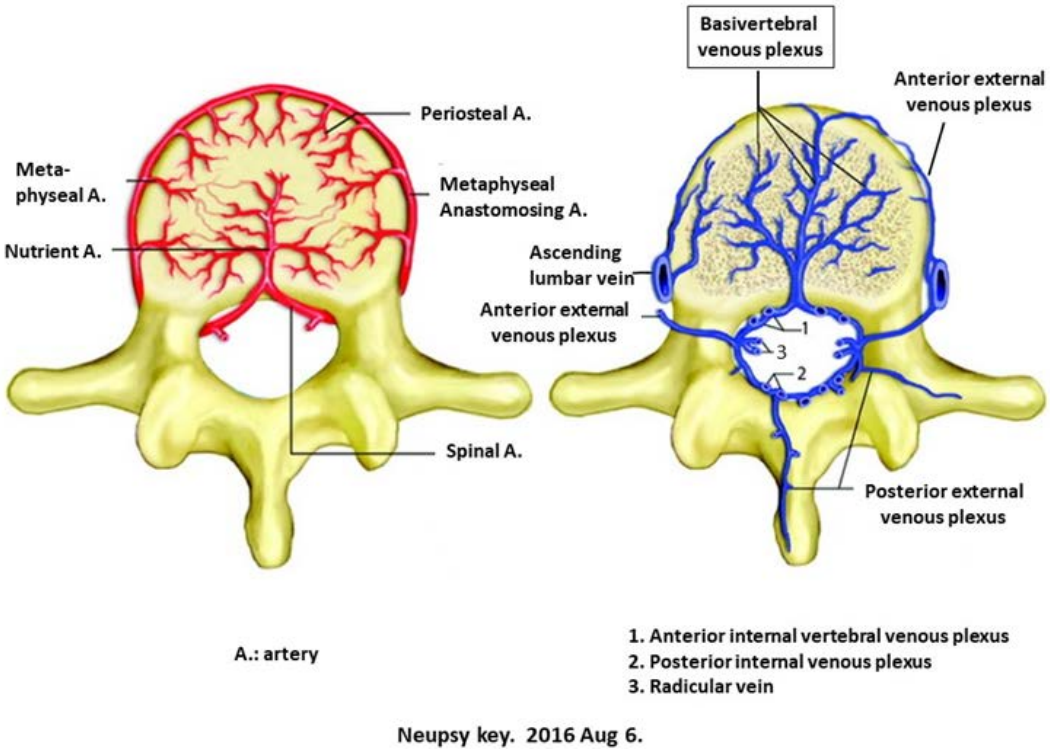

Figure 3. Vascular system of the vertebral body [7] (The figure shows the normal artererial and venous anatomy of the vertebral body)

4. Conclusion

This is the first report of hypoparathyroidism with cartwheel-like change in the thoracic vertebral body. The further accumulation of cases is necessary to determine whether this change is specific to hypoparathyroidism.

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