CASE REPORT

Multiple intracranial calcifications and spinal compressions: Rare complications of type la pseudohypoparathyroidism

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Abstract. Type la pseudohypoparathyroidism (PHP la) is an unusual inherited disease. PHP la often causes extraskeletal calcifications and even soft tissue ossifications. Patients may present neurologic symptoms and signs related to hypocalcemia and hyperphosphatemia. We report here a 38-yr-old woman with PHP la who had two uncommon neurologic complications. One was involuntary movements related to basal ganglia calcification, and the other was myelopathy owing to ossifications of the posterior longitudinal ligament and multiple herniated intervertebral disks. Aggressive body weight control and corrections of hypocalcemia, hyperphosphatemia, and elevated PTH may be important to prevent these unusual neurologic complications. Regular and careful neurologic examinations should be performed for early diagnosis and treatments of these spinal lesions.


Introduction

Pseudohypoparathyroidism (PHP) is an unusual inherited endocrine disorder. PHP was first described by Albright in 1942 (1). Five subtypes of PHP are reported, among them type la (PHP la) is the most common. The characteristic phenotype of PHP la is Albright’s hereditary osteodystropathy (AHO), which includes round face, short stature, obesity, dental hypoplasia, brachymetacarpals, brachymetatarsals and soft tissue calcification/ossification. PHP la is caused by an autosomal mutation of the α subunit of stimulating G protein (Gsα) which leads to a resistance to the PTH, resulting in hypocalcemia, hyperphosphatemia and secondary hyperparathyroidism (2). The usual neurologic complaints of PHP la, such as epilepsy, tentany and unexplained falls, are usually caused by hypocalcemia (3). Other progressive neurologic problems are rarely reported. We present here a patient with progressive neurologic complaints which were caused by intracranial calcifications, as well as cervical and thoracic myelopathies owing to ossifications of the posterior longitudinal ligament (OPLL) and herniated intervertebral disks (HIVD).

Case Report

A 38-yr-old Chinese woman was noticed to fall down frequently since the age of 2. She could walk well after calcium supplement. Mental retardation was also noted during her school years. She was asked to be given an artificial abortion by her mother at the age of 25 after an episode of hypocalcemia crisis. Since then she often had hypocalcemic attacks manifested by paresthesia, tentany and muscle twitching. During hospitalization for a suicide attempt at the age of 31, hypocalcemia (serum total calcium 2.07 mmol/l; normal range: 2.12-2.6), hyperphosphatemia (serum phosphorus 1.8 mmol/l; normal range: 0.87-1.45) and elevated serum intact PTH

Key-words: Pseudohypoparathyroidism la, spinal cord compression, spinal stenosis, multiple intracranial calcifications, ossification of the posterior longitudinal ligament.

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Because of progressive dysarthria, unsteady gait, generalized weakness, urinary incontinence and involuntary movement with twitching of both upper limbs since late 2003, she was admitted for neurologic studies in February 2004.

On admission, her height was 147.8 cm and her weight was 77.8 kg [body mass index, (BMI)]: 35.61 kg/m²). She had a typical AHO appearance, including brachydactyly of hands and feet (Fig. 1), short stature, round face, short thick neck and central obesity. The neurological examinations of cranial nerves were intact. Both Chvostek’s and Trousseau’s signs were negative. She had impaired muscle power of all limbs, especially lower extremities. Her deep tendon reflexes of bilateral knees and ankles were hyperactive. The Babinski signs were bilateral extensor plantar responses. Hypoesthesia in pinprick, temperature and vibration were noted in both feet. A wide-base gait with unsteadiness was also noted, and she could not perform a tandem gait.

The laboratory examinations revealed normal serum calcium level (2.4 mmol/l), high-normal phosphate level (1.36 mmol/l), and high-normal iPTH (52.9 pg/ml) on admission and with continuous calcitriol and calcium carbonate replacement. The serum levels of creatine, albumin, alkaline phosphatase (152 U/l; normal range: 60-220), and other electrolytes were within normal limits. The audiometry revealed no hearing loss. The cranial computed tomography (CT) showed symmetric calcifications in bilateral basal ganglia, thalamus, dentate nucleus, cerebellar and cerebral hemispheres (Fig. 2). The scalp somatosensory evoked potentials (SSEPs) showed absence of responses from each peroneal nerve and mildly prolonged central conducting times from each median nerve. An areflex neurogenic bladder without bladder sensation, bulbocavernous reflex

Fig. 1 - Radiographies show: A) sc calcifications and shorter second metatarsal bones of the right hand; and B) sc calcifications as well as shorter third and fourth metatarsal bones of the left foot.

Fig. 2 - Computed tomography (CT) of the head reveals bilateral and symmetrical intracranial calcifications in the basal ganglia, thalamus, dentate nuclei, and at gray/white matter junctions in the cerebral lobes (A) and in the cerebella (B).