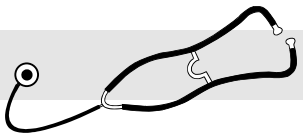

Pericardial Effusion Accompanying Isolated Hereditary Hypoparathyroidism

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Hypoparathyroidism is often a diagnostic and therapeutic challenge. Isolated hereditary hypoparathyroidism, with less than 100 cases reported, is an extremely rare disease that is often misdiagnosed. Cardiac involvement is a rare complication of isolated hereditary hypoparathyroidism, and reported cases describe cardiomyopathy and/ or congestive heart failure [1], probably related to hypocalcemia. The patient described here had a pericardial effusion. *To the best of our knowledge, this complication of the disease has not been previously reported.*

Case Description

A 54-year-old Caucasian female was admitted to the emergency room with seizures. She had been well up to 6 months earlier, when the generalized seizures began. A diagnosis of epilepsy was made and treatment with phenytoin (diphenyl-hydantoin) was initiated without improvement. At that time patchy alopecia and onychomycosis developed. One month later accelerated blindness and a cataract were diagnosed. There was no family history of any relevant diseases.

On admission, the patient appeared sick and depressed. Vital signs were normal. No lymphadenopathy was present but onychomycosis was noted. The head was normal, except for patchy alopecia. On ophthalmologic examination, bilateral cataracts with complete blindness were found. There were no skeletal deformities. The lungs were clear, and the heart sounds were distant with no murmurs. The abdomen was soft and non-tender, and the liver and spleen were not palpable. A neurological examination revealed carpopedal spasm and a positive Trousseau sign.

A complete blood count was normal. The blood chemistry profile revealed severe hypocalcemia (3.8 mg/dl) and hyperphosphatemia (7.9 mg/dl). The rest of the chemistry panel, including other electrolytes, and liver function tests were normal. Vitamin B12 levels were 347 pg/ml and folic acid 4.2 ng/ml. Antinuclear antibody and rheumatoid factor were negative. C3 measured 118 mg/dl and C4 29 mg/dl. Cortisol level was 16.6 µg/dl and the ACTH stimulation test was normal. Thyroid-stimulating hormone level was 1.0 µU/ml and parathyroid hormone 2 pg/ml (normal value 10–55 pg/ml). Serum concentrations of 1,25 dihydroxy vitamin D and 25 hydroxy vitamin D were 35 pg/ml (normal 35–50 pg/ml) and 13.4 ng/ml (normal 20–45 ng/ml) respectively. Serological tests for cytomegalovirus and Epstein-Barr virus were negative. The immunoelectrophoresis was normal.

A chest X-ray showed cardiomegaly and clear lungs. Electrocardiography demonstrated a normal sinus rhythm with low voltage and a prolonged QT interval. Ziehl-Neelsen stains of stomach aspirate and urine were negative. The small intestine barium passage failed to show evidence of malabsorption. Skeletal X-rays, including the skull, were normal. Computerized tomography of the brain and ultrasound of the parathyroid and thyroid glands were normal. Echocardiography showed a moderate pericardial effusion with normal cardiac function and morphology.

A diagnosis of hypoparathyroidism was made, phenytoin was discontinued, and intravenous calcium gluconate 10% (20 ml over 10 min, followed by infusion of 1 mg/kg/h) and oral vitamin D3 (1 µg/day) were initiated. The seizures ceased immediately and never recurred. After one day of therapy the ECG returned to normal. Over the next few weeks, the alopecia, onychomycosis and depression all disappeared. During the first month of treatment the patient began to identify shadows; a bilateral cataract extraction was performed and her vision returned to normal. The pericardial effusion disappeared gradually within 3 months.

Despite the dramatic clinical improvement, the calcium level dropped each time the patient was switched from intravenous to oral therapy (calcium carbonate 3 g/day). Once thiazide diuretics (hydrochlorothiazide 50 mg/day) were added, normal plasma calcium and phosphate levels were maintained. The patient was discharged and is doing well.

Comment

The patient under discussion presented with clinical and laboratory features suggestive of hypoparathyroidism. Since the plasma PTH level was extremely low, this case should be included in the PTH-deficient category [2]. This category includes acquired hypoparathyroidism and hypomagnesemia, both of which we excluded. There was no history of hypoparathyroidism consanguine, and the possibility of a complex autoimmune syndrome was refuted, as was a congenital defect of the parathyroid glands. The patient was thus given a diagnosis of isolated hereditary hypoparathyroidism, an extremely rare syndrome. Isolated hereditary hypoparathyroidism has been identified in autosomal dominant, autosomal recessive and X-linked inheritance patterns [3], but sporadic cases have been described as well. This sporadic form may occur at any age, but the majority of cases occur under the age of 15 [4].

The most interesting aspect of this case was the presence of a pericardial effusion, a previously unreported feature of the syndrome. In addition, hypocalcemia in itself has not been reported to cause pericarditis or pericardial effusion. Although phenytoin may theoretically cause a pericardial effusion by inducing a lupus-like syndrome, this possibility was excluded by laboratory investigation. Several cases of serositis accompanying autoimmune endocrinopathy have been reported [5], but none of these patients suffered from autoimmune hypoparathyroidism. All other efforts to elucidate the etiology of the pericardial effusion in our patient revealed no alternative diagnoses. It is of note that the pericardial effusion resolved during replacement therapy. This leads us to believe that a pericardial effusion may accompany the syndrome of isolated hereditary hypoparathyroidism. Further study is needed in order to elucidate the mechanism and assess the incidence of pericardial effusion complicating this rare disorder.

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