Brief Communication

Visual disturbances as a presenting feature of pseudohypoparathyroidism

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ABSTRACT

Introduction: Visual disturbance as a presenting feature of pseudohypoparathyroidism (PHP) is uncommon. Although papilledema is commonly reported with hypoparathyroidism primary or secondary, but not reported commonly with PHP. **Description of the Case:** A 10-year-old male child presented to our outpatient service with the complaints of blurring of vision, diplopia, and associated headache. There was no history of seizure episode. Patient had rounded face with a short, stocky built. Shortening of the fourth metacarpal and fifth metatarsal was present. Pitted nails and bilateral cataract. Patient also had clinical signs and biochemical parameters of hypocalcemia, along with normal parathyroid hormone (PTH) levels. Consistent with pseudohypopathyroidism. **Conclusion:** In cases of chronic papilledema, the assessment of the calcium serum level is a safe and simple method to exclude hypoparathyroidism or PHP.

Key words: Pseudohypoparathyroidism, papilledema, AHO Phenotype

Introduction

Pseudohypoparathyroidism (PHP) is an inherited metabolic disorder characterized by end organ resistance to the action of parathyroid hormone (PTH). PTH maintains serum calcium levels by promoting bone resorption, enhanced distal tubular reabsorption of calcium, and increased synthesis of 1,25-dihydroxyvitamin D, thereby causing enhanced intestinal calcium absorption. Resistance to PTH, therefore leads to hypocalcemia and hyperphophatemia.[1] Chronic hypocalcemia is responsible for increase in intracranial tension, thus causing papilledema, hence visual disturbances.[2]

Description of the Case

A 10-year-old male child presented to our outpatient service with the complaints of blurring of vision, double vision, and dimness of vision. History of headache was present. Patient also had history of cramping pain of extremities with spasms more in lower limb and calf muscles.

On clinical examination patient had rounded face with a short, stocky built. There was brachydactyly. Shortening of the fourth metacarpal and fifth metatarsal was present. Pitted nails and broad thumb. Bilateral cataract was present and fundus was showing presence of papilledema [Figure 1]. Trousseau's sign was positive suggestive of hypocalcemia. Findings were consistent with pseudohypopathyroidism with Albright's hereditary osteodystrophy (AHO) phenotype.[3]

Biochemical analysis is shown in Table 1. In presence of clinical features and biochemical analysis showing hypocalcemia, [Table 1] serum parathyroid levels were measured. Serum PTH value was 38.6 pg/ml which was normal. Urinary calcium was 3.57 mg/dl and urinary creatinine was 3.77 mg/dl, thus indicating renal tubular calcium loss.

In presence of hypocalcemia, renal tubular calcium loss and normal PTH and AHO phenotype, patient was labeled as a case of PHP.

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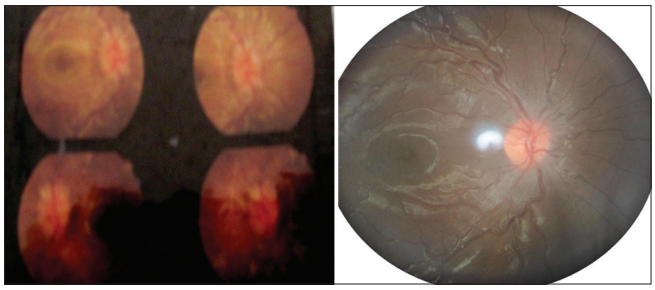

Figure 1: Fundus photograph of the patient before and after treatment

Table 1: Serum biochemistry of the patient

Parameter	31-12-2012	01-01-2013	06-02-2013	28-03-2013 (Post treatment)
Serum calcium (mg%)	7.5	7.8	7.4	9.6
Serum albumin (g%)	4.21			
Serum phosphorous (mg%)	6.1	6.0	5.9	5.2

Features of raised intracranial tension were resolved once patient was started on treatment with oral calcium, phosphate binders along with active vitamin D_3.

Discussion

PHP is called "pseudo" hypoparathyroidism because the blood chemistry values (low to normal serum calcium and high serum phosphorous) resemble the more common disorder hypoparathyroidism in which PTH is absent or low, but PTH is actually normal or elevated because the kidney cannot fully respond to PTH.[4]

A multifactorial etiology for bilateral disc edema can be postulated, such as impaired axoplasmic conduction due to reduced calcium and stasis, impaired central nervous system (CNS) vascular autoregulation causing venous stasis, and raised intracranial pressure due to pseudotumor cerebri.[2] The papilledema in hypocalcemia does not behave like that due to intracranial space occupying lesion or cerebral venous thrombosis. The disc edema readily responds to calcium therapy and disappears with calcium levels higher than 8 mg/dl.[5] Our patient also improved with normal fundoscopic findings [Figure 1] and clinical and biochemical [Table 1] improvement.

Conclusion

In cases of chronic papilledema, the assessment of the calcium serum level is a safe and simple method to exclude hypoparathyroidism or PHP.

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