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Case Report

Hypoparathyroidism: a rare cause of rickets seen in 7 years old male child

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ABSTRACT

Hypoparathyroidism is a rare condition which may occur either as an isolated disorder or in association with other autoimmune disorders. The incidence of hypoparathyroidism in the paediatric population is approximately 0.02%. Hypoparathyroidism should be kept in mind whenever a patient presents with late onset neonatal hypocalcemia or persistent hypocalcemia. Parathormone is important in maintaining calcium and phosphorus homeostasis in the body through its action on bone tissue and kidneys. Hypoparathyroidism could be either due to decreased production of the hormone or decreased peripheral action of the hormone on its target organs. Here, we present a case of a 7 years old male child who presented with complaints of bilateral lower limbs pain involving bilateral knee joints from the past 3 months along with short stature. After appropriate diagnostic work up, diagnosed as a case of rickets secondary to isolated hypoparathyroidism with short stature and started on 1-alfacalcidol 0.5 microgram and oral calcium at 100 ml/kg in two divided doses. On subsequent follow ups, patient responded to the therapy.

Keywords: Hypoparathyroidism, Rickets, 1-alfacalcidol, Autoimmune disorders, Hypocalcemia

INTRODUCTION

Parathyroid is an 82 amino acid hormone which is produced by the parathyroid glands, there are four small parathyroid glands situated behind the thyroid gland in the neck. It is crucial in maintaining calcium and phosphorus in balance in the body. Hypoparathyroidism occurs when parathyroid glands are not synthesising enough parathyroid hormone (PTH). Deficiency of the hormone results in abnormally low calcium in the blood (hypocalcemia) and high levels of phosphorus in the blood whereas excessive PTH (hyperparathyroidism) levels leads to elevated calcium levels and low levels of phosphorus in the blood. There is a reciprocal relationship between the calcium and phosphorus concentrations in the blood which is important in maintaining the integrity of bones. Calcium and phosphorus homeostasis are maintained by the influence of PTH on bone tissue and on the kidney. Hypoparathyroidism is a clinical disorder that manifests when insufficient PTH is produced or there is decreased peripheral action of the hormone on its target organs such as kidneys and bone tissue.[1] Either of these can result in hypocalcemia, which is primarily responsible for the production of the clinical features such as seizures, tetany, paraesthesias or muscle cramps, brittle nails, dry coarse skin, fatigue, weakness, patch hair loss and rickets.[2,3] Factors causing hypoparathyroidism could be any of the following such as hereditary hypoparathyroidism, autoimmune disorders, neck surgery, hypomagnesemia, extensive cancer radiation to your face or neck. PTH levels in the children with active rickets ranged from a mean of 99 pg/ml to 269 pg/ml (normal range from 9-65 pg/ml) whereas in our case it was 1.2 pg/ml, therefore clinching to a diagnosis of rickets secondary to hypoparathyroidism.

CASE REPORT

A 7 years old male child born out of non-consanguineous marriage was brought to our hospital with complaints of bilateral lower limbs pain involving knee joints which led to decreased recreational activities as there was also pain present while walking from the past 3 months and therefore, the child prefers to sit and play. The mother also complained that the child was not gaining height and was shorter as compared with his peers of the same age and sex group and also child complained of tingling sensations in the limbs. No significant antenatal or natal history and exclusively breast fed till 6 months. His review of systems was negative for emesis, diarrhoea, polyuria/oliguria, fever, appetite changes, swallowing abnormalities, respiratory symptoms, apnea, repeated acute illnesses or frequent injuries.

On examination, his age and sex appropriate height was below 3rd centile whereas both parents were of normal stature, frontal bossing, wrist widening and double malleoli and slight bowing of lower limbs were present. Developmentally normal child with neurological and other systems including genitourinary, gastrointestinal, cardiac and respiratory systems within normal limit.

There was no similar or other significant complaint in other siblings or in any other family members.

Blood investigations were done in which serum phosphorus levels were raised 8.7 mg/dl, serum 25-hydroxy vit D levels were reduced 4.29 mg/dl, serum calcium levels were reduced 6.54 mg/dl and serum parathormone were reduced 1.20 pg/ml and raised serum alkaline phosphatase levels 808.7 U/l with normal serum magnesium levels. Other laboratory works up such as complete blood count, thyroid function test, renal and liver function test were within normal limits.

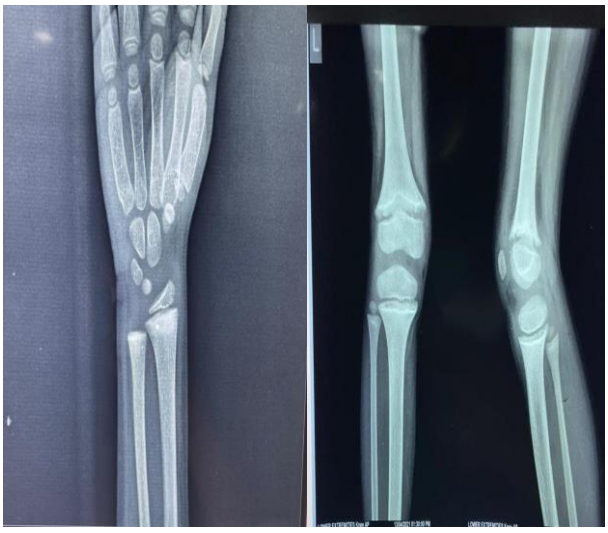

Figure 1: X-ray (wrist and knee joint) of the patient during follow up. X-ray shows the zone of calcification.

X-ray bilateral lower limbs were done showing fraying, splaying and cupping of distal end of femur and proximal end of tibia with widening of growth plate suggestive of rickets.

Subsequently, the child was diagnosed as a case of hypoparathyroid rickets and was started on 1-alfacalcidol 0.5 mcg and oral calcium BD.

Patient was called for follow ups and responded to the therapy, the child started to gain height and improvement in the bilateral lower limbs, also started to move around.

DISCUSSION

Idiopathic hypoparathyroidism, where the defined cause of hypoparathyroidism is not there is a rare condition, could be congenital or acquired later in life.

Congenital hypoparathyroidism occured during the first few months of life could be temporary or permanent. Sometimes spontaneous resolution may also occur, cause was not known. If not then usually manifested by the age of 24 months.

Most patients with congenital hypoparathyroidism had no positive family history. Cause could be absent parathyroid glands or abnormal genes that may: encode abnormal forms of PTH or its receptor, prevent normal conduction of cell signals from the PTH receptor to the nucleus, prevent normal gland development before birth.

Acquired hypoparathyroidism occured because the immune system had formed antibodies against the parathyroid gland, it can occur either in isolation or in association with other syndromes.

Hypomagnesemia should also be ruled out as it impaired the release of PTH and induces resistance to the effects of the hormone, if uncorrected.[4]

Parathyroid deficiency: hypoparathyroidism can be nonfamilial or familial and the latter is often associated with other disorders and anomalies like DiGeorge syndrome, sensorineural deafness, dysmorphic features and autoimmune polyglandular disease type I. The non-familial or sporadic variety can be transient or permanent, with the former presenting either as early as late onset neonatal hypocalcemia. The permanent variety was either isolated or associated with DiGeorge syndrome.[4] Pseudohypoparathyroidism is another clinical entity which is undifferentiated from hypoparathyroidism clinically, also presents low calcium levels and high phosphorus levels in the blood and has to be ruled out in a case of persistent hypocalcemia. In pseudohypoparathyroidism, parathyroid hormone is present but bones and kidneys do not respond to it. A short fourth metacarpal is a very useful clinical and radiological sign of pseudohypoparathyroidism; hence an X-ray wrist is advised to diagnose this condition.

Chronic patients may present with muscle cramps, pseudo papilledema, extrapyramidal signs, mental retardation and personality disturbances as well as cataracts, dry rough skin, coarse brittle hair, alopecia and abnormal dentition.

Hypoparathyroidism may lead to various complications both reversible and irreversible. Reversible complications include seizures, malformed dentition, problems with renal function, heart arrhythmia, muscle cramps. Irreversible complications include stunted growth, cataracts and slow mental development in the children.

The biochemical hallmarks of hypoparathyroidism are hypocalcemia and hyperphosphatemia in the presence of normal renal function. Serum concentrations of 1,25(OH)2 D3 are usually low and serum alkaline phosphatase is normal. Irrespective of the aetiology, the goal of the therapy was to restore normal calcium and phosphorus levels in the blood.

Vitamin D2 is the commonly used vitamin D preparation as it is the least expensive and easily available but the disadvantage is its prolonged onset of action (around 3-4 weeks) which can result in toxicity. The initial dose was 50 microgram/kg/day (2000 U/kg/day) which can be gradually stepped up so as to maintain serum calcium between 8-8.5 mg/dl. 1,25-dihydroxyvitamin D3 (Rocaltrol) has become the therapy of choice for many physicians due to its shorter half-life and a rapid onset of action (2-3 days) and metabolism, though it was costly. Therapy was initiated at 0.03 µg/kg/day, not exceeding a daily dose of 2 µg.[3,5,6]

In an emergency or acute crisis, intravenous calcium gluconate infusion at 1-2 ml/kg slowly over 30 minutes was given followed by administration of vitamin D along with oral calcium that was 100 mg/kg elemental calcium in divided doses given.[5-7]

In our case we started the child on 1-alfacalcidol at 0.5 microgram and oral calcium at 100 ml/kg in two divided doses.

CONCLUSION

Our patient was a case of rickets secondary to isolated hypoparathyroidism with short stature and started on 1-alfacalcidol and oral calcium. Hypoparathyroidism, is a rare cause of rickets. If diagnosed and treated early irreversible complications of hypoparathyroidism can be prevented. When one comes across a case of rickets, we should keep in mind every possible cause of rickets and treat promptly.

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