# Infantile Pseudohypoparathyroidism

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***Abstract-*** A case of 35 day old infant is presented who had late onset seizure associated with hypocalcemia, hyperphosphatemia, and raised parathyroid hormone. The infant did not have any stigmata of pseudohypoparathyroidism. The hypocalcemia was initially resistant to calcium therapy, but responded to vitamin D analog therapy. The diagnosis of 'pseudohypoparathyroidism' was entertained.

***Index Terms-*** Pseudohypoparathyroidism; Hypocalcaemic convulsions

## I. INTRODUCTION

Pseudohypoparathyroidism (PHP) is a heterogeneous group of disorders characterized by hypocalcemia, hyperphosphatemia, increased serum concentration of parathyroid hormone (PTH), and insensitivity to the biological activity of PTH.[1] A Medline search revealed very few case reports of PHP presenting as late onset hypocalcemia. We report a case of pseudohypoparathyroidism that presented in infantile period with hypocalcemic convulsions.

## II. CASE REPORT

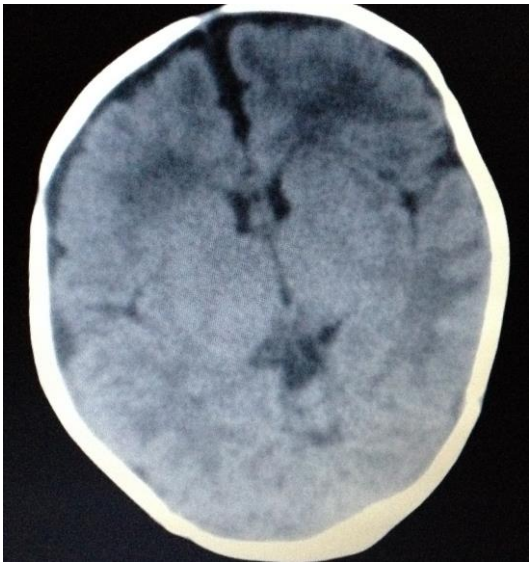
A thirty five-day-old male infant presented with a history of multiple episodes of clonic convulsions on thirty fifth day of life. The child, born to a 23-years-old multigravida at term, Birth weight (2.75 kg) cried soon after birth and was on exclusive breast feeds. The parents were healthy with normal stature and with history of third degree consanguinity. Antenatal period was uneventful.

The baby when admitted weighed 3.715 kg, with head circumference of 37 cm. Anterior fontanelle was at level and there was no dysmorphic facies or gross congenital malformation. Rest of the systemic examination including ophthalmological examination was normal.

Provisional diagnosis of metabolic seizure was made and investigations revealed blood glucose 90 mg/dL and Blood urea was 24 mg/ dL, Sr.creatinine was 0.4mg/dl,Sr.Albumin was 3.5g/dlLab parameters revealed hypocalcemia,hyperphosphatemia, raised parathormone ,Vitamin D level was not deficient. The values of calcium,phosphorus,magnesium,ALP, parathormone and Vitamin D are shown in the adjacent table Other electrolytes were within normal limits and septic screen was negative. Ultrasound skull and abdomen did not reveal any abnormality. X-ray chest was normal.ECG showed prolonged QT interval with no arrhythmia Thyroid profile was normal. Child was started on I.V.Calcium gluconate and repeat calcium and phosphorous was done after 3 days which revealed Sr.Calcium(6.8mg/dl),Sr.Phosphorus(8mg/dl)

| **Serum Metabolites** | **Values** mg/dl | **Reference range** mg/dl |
|---|---|---|
| Calcium | 5.4 | 8.4-10.2 |
| Phosphorus | 11.0 | 3.8-6.5 |
| Magnesium | 2.8 | 1.6-2.3 |
| Alkaline phosphatase | 572 U/L | 150-400 U/L |
| Parathormone | 138.7 pg/ml | 14-72 pg/ml |
| Vitamin D | 30.56 nmol/L | <25 nmol/L is deficient |

Sr.Calcium and Sr.Phosphorous and Alkaline Phosphatase of mother were normal.In view of persistent hypocalemia, hyperphosphatemia and high serum parathormone levels, diagnosis of Pseudohypoparathyroidism was made, and the child was treated with calcium supplementation and calcitriol 0.25 µg/day. Child was discharged and advised to come for follow up.



***CT Brain Showing Normal Study.***

## III. DISCUSSION

In 1942, Fuller Albright first introduced the term pseudohypoparathyroidism to describe patients who presented with PTH-resistant hypocalcemia and hyperphosphatemia. In PHP, the parathyroid glands are normal or hyperplastic histologically, and neither endogenous nor administered PTH raises the serum levels of calcium or lowers the level of phosphorus.

Pseudohypoparathyroidism is divided into 2 main types. Type I is characterized by low or absent renal cyclic adenosine monophosphate (CAMP) production in response to parathormone (PTH). Type II responds to PTH with normal increase in urinary CAMP but shows absent or subnormal phosphaturic response[2]. Type I is further subdivided into 2 subtypes, A and B. In sub type A, the affected patients have a genetic defect of the a subunit of the stimulatory guanine nucleotide binding protein (GSα), with most of them having distinctive morphological abnormalities collectively called "Albrights herediatry osteodystrophy".[1] In this type, hypocalcemia rarely develops before 3 years.[3] Subtype I B patients have normal levels of G protein activity with defect in PTH receptor expression or a defect in catalytic subunit of adenyl cyclase.

In the present case, the infant presented with hypocalcemic convulsions. Hypomagnesemia, septicemia, renal failure were ruled out. There were no predisposing factors for hypocalcemia like prematurity, birth asphyxia. Elevated levels of serum parathormone levels further ruled out hypoparathyroidism. A case of four day old neonate with pseudohypoparathyroidism has been reported previously in 2006[7]. Other cases reported were mostly of 8 -13 year old age group.

Since this infant presented at 35 day of life, we are reporting this case due to its rarity The child is on oral calcium supplementation and calcitriol and is seizure free and under follow up. All patients with severe symptomatic hypocalcemia should be initially treated with intravenous calcium. Administration of oral calcium and 1 alpha hydroxylated vitamin D metabolites, such as calcitriol, remains the mainstay of treatment and should be initiated in every patient with a diagnosis of PHP. The goals of therapy are to maintain serum total and ionized calcium levels within the reference range to avoid hypercalciuria and to suppress PTH levels to normal. This is important because elevated PTH levels in patients with PHP could cause increased bone remodeling and can lead to hyperparathyroid bone disease. To conclude any child presenting with late onset hypocalcemic seizure, parathormone levels are to be checked along with Vitamin D.

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