

## Case Report

# Congenital Hypoparathyroidism Presenting with Stridor and Severe Growth Failure in an Infant: A Rare Case

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### Abstract:

**Background:** Congenital hypoparathyroidism is a rare endocrine disorder characterized by hypocalcemia due to deficient secretion or action of parathyroid hormone. The clinical manifestations of this disorder are variable in nature and clinical presentation; however, the most common signs/symptoms of symptomatic cases include seizures, tetany and muscle twitching. The presence of stridor secondary to a laryngospasm as a presentation of congenital hypoparathyroidism is uncommon and frequently missed, particularly when other neurological problems do not exist.

**Case Presentation:** A six-month-old female infant, born via lower uterine segment cesarean section at 35 weeks gestation to a diabetic mother, presented to our hospital with severe growth failure (5 kg), feeding difficulties, respiratory distress and stridor. Her neonatal period was complicated by respiratory distress and MRSA sepsis, which required admission to a neonatal ICU. After that, she was managed for gastroesophageal reflux and a small atrial septal defect, but had persistent difficulties with feeding and breathing. When she was admitted to our unit, she had no evidence of lower respiratory disease; her chest radiograph was normal. Laboratory results revealed significant hypocalcemia, very low levels of serum PTH and normal levels of vitamin D, thus confirming the diagnosis of congenital hypoparathyroidism.

**Conclusion:** This case highlights a rare presentation of congenital hypoparathyroidism with stridor and severe growth failure without seizures. Clinicians should consider hypocalcemia in infants presenting with unexplained stridor and feeding difficulties. Early diagnosis and appropriate management are essential to prevent complications and improve clinical outcomes.

**Keywords:** Congenital hypoparathyroidism, Hypocalcemia, Stridor, Laryngospasm, Infant, Growth failure.

### Introduction:

Hypoparathyroidism in children is a rare endocrine disorder and is usually asymptomatic.<sup>1</sup> Congenital

hypoparathyroidism is characterized by hypocalcemia secondary to impaired production, action, or secretion of

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parathyroid hormone (PTH) and is a heterogeneous disorder.<sup>2</sup> Serum PTH remains low throughout life in familial hypoparathyroidism.<sup>2</sup> Few patients have symptomatic hypocalcemia, including lethargy, recurrent

respiratory tract infections, focal clonic seizures, jitteriness, carpopedal spasm and rarely stridor due to laryngospasm.<sup>3</sup> Stridor occurring as a consequence of nutritional rickets, which is reported in the literature.<sup>3</sup> Hypocalcemia has also been shown to occur in exclusively breast-fed babies not exposed to sunlight and in infants whose mothers are deficient in vitamin D.<sup>4</sup> Our baby presented with respiratory distress and stridor with severe growth failure and feeding difficulty without any convulsion, which is a rare presentation in infancy.

**Case Report:**

Our baby girl was delivered at 35 weeks of gestational age, weighing 2500 g, by LUCS as the 4th issue of a diabetic mother (age 34 years) due to less fetal movement and uncontrolled diabetes. After birth, she was in the NICU for 10 days for respiratory distress and sepsis (MRSA). From the 3rd week of life, she developed frequent vomiting after feeding and was diagnosed with GERD. Due to recurrent respiratory complaints, an echo was done, and a small ASD was discovered at 1 month of age. For these issues, she was treated with domperidone, frusemide, potassium, and vitamins. But her mother always complained of abnormal respiration and feeding difficulties, which occurred at home but were not seen during follow-up. At the age of 6 months (Figure 1), she was admitted to the hospital with a weight of 5 kg, a severe feeding problem associated with chest indrawing & stridor. She had no rhonchi in her chest. Her chest x-ray was normal, but she had hypocalcemia with severe low parathyroid hormone & normal vitamin D (Table 1).



**Figure 1:** Baby at 6 months of age.

**Table 1:** Laboratory findings at the age of 6 months.

Investigation	Patient's value	Reference value
Serum calcium	3.33 mg/dl	10-12mg/dl
Inorganic phosphate	7.92mg/dl	2-5 mg/dl
Vitamin (D2+D3)	34.3 ng/ml	Sufficient 30-100 ng/ml
Parathyroid hormone	3.34pg/ml	15-65 pg/ml

The baby was treated with 60 mg/kg calcium gluconate, 200 IU vitamin D and folic acid, and an iron supplement as required. The baby was discharged with the above medication, but when the patient took oral calcium in tablet form, stridor appeared again. So, she kept on injecting calcium mixed with milk. At her 9 months of age (Figure 2), 2 investigations were done. (Table 2).



**Figure 2:** Baby at 9 months of age.

**Table 2:** Laboratory findings at the age of 9 months.

Investigation	Patient's value	Reference value
Serum calcium	6.05 mg/dl	10-12mg/dl
Parathyroid hormone	2.06pg/ml	15-65 pg/ml

Now the baby girl turns 1 year old, but the parents are least interested in doing an investigation. She has gained weight only 7 kg, but development is near age-appropriate, like walking with support and speaking some words.

**Discussion:**

Hypoparathyroidism is a rare condition in which the body secretes abnormally low levels of PTH.<sup>5</sup> PTH is responsible for the regulation and maintenance of calcium and phosphorus in the body. Hypoparathyroidism

can occur due to congenital hypoplasia/aplasia, which may be accompanied by other congenital anomalies, such as dysmorphic facies, immunodeficiency, lymphedema, nephropathy, nerve deafness, or cardiac malformation.<sup>2</sup> In our case, there is no facial dysmorphism; only growth failure and feeding difficulty are observed. Hypoparathyroidism represents a wide range of clinical and biochemical syndromes characterized by parathyroid hormone deficiency, hypocalcemia, and hyperphosphatemia, which is present in the reported baby.<sup>6</sup> Neonatal hypoparathyroidism is a relatively common transient condition associated with well-defined risk factors such as pre-maturity, perinatal asphyxia, and maternal diabetes.<sup>2</sup> Permanent congenital hypoparathyroidism is rare.<sup>6</sup> Mother had gestational diabetes, but the baby had no convulsions during the neonatal period. She had respiratory distress and was suffering from Transient Tachypnea of the Newborn (TTN), and incidentally, ASD was discovered.

Congenital hypoparathyroidism has been reported in association with a variety of developmental anomalies, including lymphoedema, nephropathy, nerve deafness, congenital heart disease and chromosomal abnormalities.<sup>6</sup> The high-pitched crowing sound of laryngospasm due to tetany (laryngeal stridor) is most likely in tetany of the newborn, but it may also be made in older children with increased neuromuscular instability, provoked by a state of alkalosis, whether due to celiac disease, rickets, hypoparathyroidism, renal failure or hyperventilation due to hypocalcemia.<sup>7</sup> Our baby had no such facial dysmorphism or neuromuscular problem, including rickets or renal failure. Only laryngospasm was present during the 6 months of age, which was mistaken for stridor, which is a common respiratory symptom in upper respiratory diseases like acute laryngotracheitis.<sup>7</sup>

PubMed published a review of stridor and hypocalcemia. It revealed 22 publications which were reviewed; 15 of these publications discussed the association between stridor and hypocalcemia, of which two were in elderly people, and the rest were mostly case reports in infants.<sup>7</sup> Stridor was the presenting feature in all cases, with or without convulsions, but all had hypocalcemia. This feature is similar to our reported baby.

The infant reported by Venkatesh & Chhavi was managed as a case of acute laryngotracheobronchitis initially, as there was no evidence of rickets, and the diagnosis of hypocalcemia was made only after he had convulsions. This case report highlights the importance of measuring serum calcium levels in infants with acute

stridor and or wheeze, as in our case. Management consists of correcting hypocalcemia, determining the cause of hypocalcemia, and treating it. Prompt resolution of symptoms on correction of hypocalcemia, as had happened in our case, which was reported in previous literature.<sup>3</sup>

Patients with chronic hypoparathyroidism might remain asymptomatic due to adaptation to calcium levels as low as 7.0 mg/dL, but they may also present with various symptoms ranging from mild to severe.<sup>8</sup> In newborns, hypocalcemia is associated with feeding difficulties, vomiting, lethargy, exaggerated startle, myoclonic beats, convulsion, apnea, tachypnea, tachycardia, cyanosis, laryngospasm, and heart failure.<sup>8</sup> Our baby had only laryngospasm, feeding difficulties and was less responsive.

#### Conclusion:

The aim of reporting this case is to emphasize the extremely rare presentation that mimics very common illnesses like bronchiolitis or epiglottitis. But a high index of suspicion should be kept in mind to help diagnose rare disorders like congenital primary hypoparathyroidism. The major goals for management are to prevent symptomatic hypocalcemia and to ameliorate the complications of the disease.

**Conflict of interest:** There is no conflict of interest.

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