Case Report

A Rare Case of Primary Idiopathic Hypoparathyroidism Presented with Early Infantile Seizures

Madoori Srinivas[1], Sudhakar Chiluka[2], Anitha K[3], Subhan Basha[3], Praneet Reddy N[3]

[1] Professor
[2] Assistant Professor
[3] Junior Residents
Department of Paediatrics
Chalmeda Ananda Rao
Institute of Medical Sciences
Karimnagar - 505 001
Telangana, India.

CORRESPONDENCE :

Dr. Madoori Srinivas, MD (Paediatrics)
Professor & HOD
Department of Paediatrics
Chalmeda Ananda Rao
Institute of Medical Sciences
Karimnagar - 505 001
Telangana, India.
E-mail:
madoorisrinivas@gmail.com

ABSTRACT

Seizures are common during childhood. Most common cause for seizures in children between 6 months to 5 years is febrile seizures, in school going children seizures are due to idiopathic epilepsy. Whereas etiology of infantile seizures is diverse. Infantile seizures usually due to metabolic causes such as hypoglycemia, hypocalcemia and hypomagnesaemia. These are more commonly seen in infants with perinatal asphyxia, prematurity and intrauterine growth retardation. Hypocalcemia due to primary hypoparathyroidism either transient or permanent is rare. It presents with biochemical abnormalities like hypocalcemia, hyperphosphatemia and low levels of Parathyroid hormone (PTH). We are reporting a case of 39 days old infant presented with seizures due to hypocalcaemia because of primary idiopathic Hypoparathyroidism.

Keyowords: Infant, seizures, hypocalcaemia, primary idiopathic hypoparathyroidism

INTRODUCTION

Seizures in infantile period have diverse etiology. Hypocalcemia is one of the treatable metabolic cause[1], hypocalcemia may be transient or permanent, primary or secondary. Hypoparathyroidism is one of the rare causes of hypocalcemia.

Isolated congenital (idiopathic) primary hypopara thyroidism is a rare which is usually sporadic but familial cases have also been reported,[1, 2] Hypoparathyroidism can occur as a part of maternal autoimmune disorders, polyendocrinopathy with dysmorphic syndromes like CATCH22, basal ganglia calcification, postthyroidectomy complications, Di George syndrome, Velo-cardiofacial defect1.[3-5] True incidence is not known but is reported as 7.2/million people from Japan.

CASE REPORT

A female baby born out of nonconsanguinous marriage, 3rd in birth order presented on 39th day of life with seizures, one episode per day for the last three days. First episode was inconsolable excessive cry for 10 min, second, and third episodes are generalized tonic clonic seizures (GTCS), each episode lasted for about 3-5 min. There is no history of loss of consciousness, no bowel and bladder disturbances, and no focal neurological deficits.

Seizures stopped spontaneously, in postictal and interictal period baby remained normal. There is no history of fever, vomiting, diarrhea, head injury, or ear discharge. The baby was delivered by cesarean section; indication for section is previous section, cried immediately after birth, appropriate for gestation age, no neonatal problems. Infant was on exclusive breast feeding. There is a history of second sibling female infant died at the age of 35 days due to seizures; first sibling is male 4 years, healthy, no history of maternal illness or drug intake during pregnancy.

On examination baby was irritable, anterior fontanella patent, afebrile, heart rate and respiratory rate were within normal limits, head circumference, length and

weight at 50th centile, no dysmorphic features were seen. Systemic examination was normal. Baby was admitted in paediatric intensive care unit of Chalmeda Ananda Rao Institute of Medical Sciences, Karimnagar and secured intravenous line, sent baseline investigations, as per ICU protocol in view of multiple episodes of seizures intravenous Phenobarbitone 20 mg/kg given slowly.

Investigations revealed normal blood counts, blood glucose, electrolytes, C reactive protein, and renal function tests. Serum Calcium 6.5mg/dl was noted from the investigations done at the time of presentation considered as hypocalcemic seizures and treated with intravenous calcium (Table 1 & 2).

Baby remained well for 22 hours and again had a seizure activity which was managed with IV Midazolam. Repeat testing of calcium, blood glucose, electrolytes, and magnesium was done repeat investigations again revealed hypocalcemia even after iv calcium supplementation.

Further laboratory workup to rule out conditions causing hypocalcemia which included ABG, chest x-ray, neurosonogram, phosphate, Vit.D, Parathyroid hormone was done which were suggestive of hypoparathyroidism (decreased calcium, PTH, and increased phosphate) (Table 2).

X-Ray chest and Echocardiography were done to rule out Di George syndrome. Maternal calcium, phosphate and ALP were within normal limits. Final diagnosis of primary idiopathic hypoparathyroidism was made and treated with IV calcium and oral cholecalciferol. Seizures are controlled and discharged on oral medication (calcium and cholecalciferol) and upon follow up the baby is found to be normal.

Table: 1 Basic Investigations

Haemoglobin	13.9gm/dl
Total Leukocyte count	19400/cumm
Differential Count	Neutrophils-48%, Lymphocytes-46% Eosinophils-2% Monocytes-4%
Platelet count	7lakhs/cumm
GRBS	158mg/dl
Serum calcium	7mg/dl, Ionized Calcium-0.53mmol/l
1,25 OH Vit D3	34ng/ml
Serum Electrolytes	Sodium-139mmol/l,Potasium-6mmol/l,Chloride-100mmol/l
Blood Culture	Negative
Blood urea	17mg/dl
Serum creatinine	0.6mg/dl
LFT	Total protein-5.8gm/dl;Albumin-3.6gm/dl; Globulin-2.2gm/dl Total bilirubin-0.8mg/dl, A:G-1.6 ; SGOT- 43U/L; SGPT- 47 U/L, ALP- 251 U/L; GGT-114 U/L.
Chest -X-ray	Normal

Table 2 : Advanced Investigations

Serum Phosphorus	9.3 mg/dl
Serum Magnesium	2 mg/dl
PTH	1 pg/ml
EEG	Normal
CT brain	Normal
2D ECHO	Normal
Neurosonogram	Normal
USG Abdomen	Normal
Thyroid profile	T3-1.7 ng/ml, T4-169 ng/ml, TSH-4.9 µIU/ml
CSF Analysis	Glucose-42 mg/dl; Protein-35 mg/dl; ADA-2 U/L; LDH-283 IU/L; Colour-clear; Microscopic examination-TC-2 cells/cumm, DC-All are lymphocytes
Urine Calcium Creatinine Ratio	0.56

DISCUSSION

Hypocalcaemia is a common metabolic cause for the seizures during neonatal period where as in infantile period febrile convulsions are more common. Hypocalcemia is defined as total serum calcium of less than 7 mg/dl or ionized calcium less than 4 mg/dl. Hypoparathyroidism is a rare cause of hypocalcaemia. Usually hypocalcemia is seen in preterm, LBW babies, and rare in term infants.

Hypoparathyroidism can occur in isolation or in combination with other autoimmune/ genetic defects.[1,3-5] Sanjad and Sakati et al have described such associations of hypoparathyroidism with facial dysmorphism, growth failure, and mental retardation.[7] Di George syndrome has also association with hypocalcemia.

Isolated congenital hypoparathyroidism can occur as a sporadic or familial disorder with inheritance by autosomal dominant, recessive or X linked modes of transmission.[6] In this case death of the sibling with seizures may be due to hypoparathyroidism and can be considered for familial hypoparathyroidism, Defects in the prepro PTH gene located at 11p15 gene locus and mutations in calcium sensing receptor gene (3q 21-24) have been associated in cases of isolated PTH deficiency.[8]

One of the differential diagnoses is hypomagnesemia it can lead to decreased levels of calcium, Vit D and PTH. But if we correct hypomagnesemia with parenteral magnesium then calcium, Vit D and PTH levels are automatically corrected.

In our case patient presented on the 39th day of life with seizures and had hypocalcemia, raised phosphate, low PTH levels and normal magnesium in comparison with two cases present ed by Atika et.al[1] of isolated congenital hypoparathyroidism. Patient was treated with calcium supplementation, calcitriol and remained seizure free. Similarly Rocha et al[8] presented a case of new born with hypocalcaemia and low PTH levels but their patient had hypomagnesaemia.

CONCLUSION

Aim of reporting this case is to emphasize that, presentation of rare disorders may mimic with common illnesses but a high index of suspicion should be kept in mind to diagnose rare disorders like primary hypoparathyroidism especially healthy infant with seizures. With early diagnosis, management prognosis is good thereby complications of seizures can be avoided.

CONFLICT OF INTEREST :

The authors declared no conflict of interest.

FUNDING : None

AKNOWLEDGEMENTS:

We are very much thankful to Chairman Chalmeda Laxminarasimha Rao, Director Dr. V. Suryanarayana Reddy for permitting us to publish this case report. We are very much thankful to the parents of patient for giving consent to report.

REFERENCES

1. Atika Z, Yaqoob M, Waseem R, Abbas A. Isolated congenital hypoparathyroidism: Follow up of two sisters. *Pak Pediatr J.* 2013; 37:188-190.

2. Drake TG, Albight F, Bauer W. Chronic idiopathic hypoparathyroidism: report of 6 cases with autopsy findings in one. *Ann Intern Med.* 1934; 12:1751. doi: 10.7326/0003- 4819-12-11-1751.

3. Ramen CB. A case report of basal ganglia calcification- a rare finding of hypoparathyroidism. *Oman Med J.* 2009; 24:84-86. doi: 10.5001/omj.2009.44

4. Akçakuþ M, Güneþ T, Kurtoðlu S, Çetin N, Özkul Y, Narin N, et al. Asymmetric crying facies associated with congenital hypoparathyroidism and 22q11 deletion. *Turk J Peds.* 2004; 46:191-193.

5. Patil MB, Patil SM. Facio-Auriculo-Vertebral sequence in association with congenital hypoparathyroidism. *Ind Peds.* 2012; 49:670-671.

6. Sanjad SA, Sakati NA, Abu-Osba YK, Kaddoura R, Milner RDG. A new syndrome of congenital hypoparathyroidism, severe growth failure and dysmorphic features. *Arch Dis Child.* 1991; 66:193-196.

7. Finegold DN, Armitage MM, Galiani M, Matise TC, Pandian MR, Perry YM, et al. Preliminary localization of a gene for autosomal dominant hypoparathyroidism to chromosome 3q13. *Pediatr Res.* 1994; 36:414-417. doi: 10.1203/00006450- 199409000-00024.

8. Rocha C, Gonnetti N, Pelluci L, Rocha MS. Hypocalcemia and neonatal seizures: a rare case of congenital hypoparathyroidism. *Arq Neuro Siquitr.* 2002; 60:138-141. doi: 10.1590/s0004-282X2002000100025.