A CASE REPORT OF EARLY BIOTINDASE DEFICIENCY

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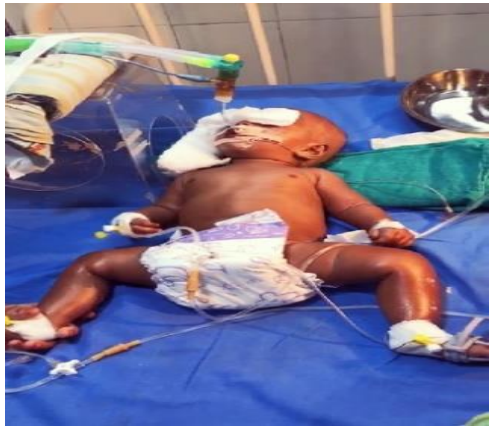
INTRODUCTION

Biotinidase is responsible for the cleavage of biotin from biocytin. Complete or partial absence of biotinidase is associated with a wide spectrum of clinical manifestations like vomiting, lethargy, hypotonia, skin rashes, seizures, developmental delay, alopecia. The seizures are typically not responsive to conventional anticonvulsant therapy, though rapidly responsive to biotin therapy.

PATIENT PRESENTATION

A 4 months old boy born out of a 3rd degree consanguinous marriage, 2nd in birth order, with birth weight 2.5kg, presented with seizures, vomiting, loss of scalp hair for 1 month. The child was normal till 3 months, then started having many episodes of generalized seizures, with each episode lasting for 3-5 min, for which child was started on syp. Phenobarbitone. The child had developmental delay in the form of not attaining social smile, not recognizing mother, no head control. No family history of epilepsy and similar complaints. On the day of admission, the child had repeated episodes of seizure without gaining consciousness between those episodes, with severe respiratory distress in the form of subcoastal and intercoastal retractions.

PICTURES



At admission

At follow-up

CLINICAL COURSE

On examination , baby was convulsing with vitals PR- 202bpm, RR- , SpO2- 80% with 2 litres 02 with nasal prongs. CRT < 3 seconds, Pallor was present. No bulging of anterior fontanelle.

INVESTIGATIONS

Investigations revealed Hb- 8.8 gm%, GRBS - 117 mg/dL, Chest X-ray was normal. LFT, RFT, Serum electrolytes were within normal limits. ABG analysis reveals severe metabolic acidosis pH 7.05, pCO2 20mmHg, HCO3 6.6 mmol/lit. TMS report was suggestive of biotinidase deficiency with biotinidase level of 24 IU (normal > 50 IU) and elevated levels of hydroxyisovaleryl carnitine.

DISCUSSION

The child was started on anticonvulsants, intubated and was on mechanical ventilation. Metabolic acidosis was corrected with sodium bicarbonate infusions. After diagnosing as early infant biotinidase deficiency, child was started on biotin 30mg/day. Seizures stopped after 3 days of starting biotin. Childs's general condition improved and was extubated after 2 more days. Childs started taking breastfeeds, doing well and hence discharged with advice to continue biotin.

CONCLUSION

Biotinidase deficiency is a classic example of vitamin responsive disease. Age of presentation depends on whether the deficiency is partial or total. Treatment is supplementation of biotin 5-20 mg/day(some may require 30-60mg/day)

REFERENCES

1.Burri BJ, Sweetman L, Nyhan WL. Mutant holocarboxylasesynthetase: evidence for the enzyme defect in early infantile biotin-responsive multiple carboxylase deficiency. J clin investigation. 1981;68: 2. Wolf B, Heard GS, Weissbecker KA, McVoy JR, Grier RE, Leshner RT. Biotindase deficiency: initial clinical features and rapid diagnosis. Ann Neurol. 1985;18:614-7.