BACKGROUND

- Peroxisomal biogenesis disorders are autosomal recessive disorders characterized by defective biosynthesis, assembly and function.
- The primary cause of these disorders is due to mutations in PEX gene.
- PBDs are classified into two types: PBD- Zellweger spectrum disorder (PBD-ZSD) and rhizomelic chondrodysplasia punctata type 1
- Hypoglycaemia is not reported in peroxisomal disorders. Although peroxisomes play a role in very long chain fatty acid oxidation, the main pathway of fatty acid oxidation (FAO) occurs in mitochondria.
- Hypoglycaemia occurring in a child with PBD needs thorough work-up to find the etiology.

CASE

- A 7 month old boy was referred with the complaints of irritability and excessive cry.
- He was known to have multiple problems in the form of bilateral sensorineural hearing loss, global developmental delay, hypotonia, visual impairment and gastroesophageal reflux.
- No family history of diabetes mellitus or hypoglycaemia was reported.
- He was born at 41 weeks of gestation with a birth weight of 3.7 kg (0.48 SDS).
- On examination hepatomegaly, dysmorphic features including asymmetry of nasoal crease, sacral dimple, metatarsus adductus and right undescended testes were noted.
- Investigations revealed deranged liver function [AST- 1576 IU/L (12-41), ALT- 918 IU/L (80-36), GGT- 183 IU/L (0-50), Total Protein- 67 g/L (67-92), Albumin- 40 g/L (38-58), PT-15.3 sec (9.1-11.8), INR- 1.48, APTT- 27.6 sec (22.8-34.7)], normal serum electrolytes and blood gas analysis.
- The child started to have repeated episodes of Hypoglycaemia requiring high glucose infusion rate (GIR) of 10.6 mg/kg/min.
- Critical sample during hypoglycaemic episode revealed a blood glucose of 2.5 mmol/L (<2.6 mmol/L), Insulin of 18 mIU/mL C-peptide of 116 pmol/L and beta hydroxyl butyrate (<100 µmol/L) and free fatty acid undetectable (FFA) (<275 µmol/L) were undetectable suggestive of HH.
- The child was started on intravenous glucagon and high concentration intravenous dextrose (GIR 10.6 mg/kg/min) to which he responded well. Echocardiogram showed a structurally and functionally normal heart study.
- The cortisol was 364 nmol/L during hypoglycaemia. Standard short synacthen test revealed a baseline cortisol of 147nmol/L and a suboptimal peak of 214nmol/L suggesting adrenal insufficiency.

CONCLUSION

- Hypoglycaemia in a child with peroxisomal disorder needs a thorough approach.
- Although rare, HH should be considered.
- Treatment with diazoxide could help in the management of hypoglycaemia.

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