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NEONATAL PERSISTENT HYPOGLYCEMIA AND HYPERGLYCEMIA (REPORT OF TWO CASES)

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Backround : Hypoglycemia and hyperglycemia are common metabolic problems, detected especially in NICU patients. Most of them are transient and related with prematurity, SGA delivery, sepsis, infants of diabetic mothers and hypoxia-ischemia. However hypo or hyperglycemia may sometimes be persistent. In these cases, rare disorders of glucose metabolism such as islet-cell hyperplasia, betacell adenomatozis, glycogen storage disease for hypoglycemia and neonatal diabetes mellitus, Schwahmann-Diamond Syndrome, Johanson-Blizzard Syndrome for hyperglycemia, should be investigated. The diagnosis of such rare disorders are difficult and some of them depend on pathologic evaluation.
Cases : We present two cases with rare disorders of glucose metabolism. The first case had persistent hypoglycemia despite high glucose infusion (25 mg/kg/min.) and administration of octreotide. The diagnosis of nesidioblastosis could be made by pathologic evalution of pancreatic tissue obtained at autopsy. The second case had persistent hyperglycemia and edema due to hypoalbuminemia. His glucose level could not be decreased despite high insulin therapy. Pancreatic agenesis was diagnosed at autopsy.

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NEONATAL NONKETOTIC HYPERGLYCINEMIA AND MR SPECTROSCOPY FINDINGS IN A NEWBORN

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Background: Neonatal nonketotic hyperglycinemia (NNH) is a metabolic disorder due to deficiency of the hepatic glycine cleavage reaction. This leads to accumulation of glycine in all body tissues and fluids with a possible fatal course.
Case: A ten-hour-old neonate with hypotonicity and failure to suck was referred to our center with suspect of metabolic disease. The infant was the second living child of the family and the parents had first degree cousin marriage. The first baby died in the first days of life without any diagnosis. Mother had an uneventful course of pregnancy with normal fetal movements. The infant was delivered by the vaginal route and there was no history of perinatal asyphxia. Failure to suck and hypotonicity developed gradually in a few hours following delivery. The infant was appropriate for gestational age and had a normal phenotype. She was lethargic, hypotonic, hypoactive with hiccups, but showed response to painful stimuli. Routine examination of cerebrospinal fluid (CSF) and complete blood counts were normal. There was no ketosis, asidosis, hypoglycemia, hyperammonemia, hepatocellular and renal disfunction. Serum and urine amino acid levels were studied. Glycine was found in high amounts in plasma (1,865 μmol/L) and urine (17,461 μmol/L). Glycine level of CSF was also increased (190.9 μmol/L) and the ratio of CSF glycine to plasma was 0.10. In electroencephalography there were burst and supression complexes. MR spectroscopy revealed agenesis of corpus callosum with neuronal loss; n-acetylaspartate amino acid signal loss in both parietooccipital white matter and cortex and also a peak in glycine signal at 3.5 ppm. In the second day, respiratory support by mechanic ventilation and fenitoin infusion for generalized myoclonic convulsions were started. The infant started spontaneous respiration on the 15th day and externated from the NICU. She had intractable convulsions and died on the 4th mounth in his second admittion to NICU with aspiration pneumonia.
Conclusion: NNH should be considered in infants with hypotonicity, intractable hiccups, convulsions, lethargy and apnea.

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