Case report

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Perinatal outcome complicated by long chain fatty acid disorder: a case report

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Introduction

Fatty acids comprise the largest energy reserve in humans, and constitute a vital role in supplying energy during instances of fasting and stress through a B oxidation pathway in the mitochondria. This metabolic pathway is critical in neonates who have a paucity of glycogen reserve and high metabolic rate; occurrence of a precipitous metabolic decompensation can occur if any disruption occurs along the enzymatic pathway. Long Chain Fatty Acid Oxidation Disorders (LC-FAODs) are a heterogenous set of rare, potentially fatal autosomal recessive genetic maladies, which can impact both the perinatal and infantile periods. These disorders can increase the risk of adverse perinatal outcomes, inclusive of preterm labor/delivery, IUGR, preeclampsia spectrum, and neonatal compromise/demise. We present a case of a perinatal outcome impacted by a LC-FAOD.[1]

Case report

A 22 year old primiparous Hispanic woman presented as a late registrant at 29 weeks gestation. Her prenatal course was complicated by no prenatal care, a language barrier and an episode of preterm contractions 1 week prior. Sonogram revealed mega cisterna magna of the fetus. Genetic evaluation/ consultation revealed low risk NIPT, and patient declined genetic amniocentesis. Patient presented at 34w6d in early labor, with marked elevation of blood pressures consistent with preeclampsia with severe features. Empiric antibiotics were given for unknown GBS status. The patient delivered vaginally 5 hours later; female infant, 2025 grams, APGARS 9/9. GBS culture obtained at admission later returned positive.

At one hour of life, hypothermia of 92.1°F was noted and the baby was placed under a warmer with noted improvement. At 32 hours of life, was again hypothermic, weak and with nasal flaring. The baby was subsequently admitted to NICU for sepsis evaluation and monitoring. CBC, CMP, CRP, blood cultures, Lumbar puncture performed; empiric antibiotics of Ampicillin and Gentamicin initiated. Sonogram of the baby's brain was negative for mega cisterna magna. At 50 hours of life, oxygen desaturations, tachypnea, bradycardia were noted; despite intubation and continuous resuscitative measures the neonate eventually expired. Pertinent findings: Elevated liver transaminase and a newborn screen revealing LC-FAOD protein deficiency, CSF analysis- pink color, low glucose, elevated protein. The NYS Newborn Screening Program report on this patient results were indicative of long-chain acyl-CoA dehydrogenase (LCHAD) deficiency or trifunctional protein (TFP) deficiency. Neonatal autopsy indicated that the *immediate* cause of death was sepsis, with involvement of the brain and the lungs; the LC-FAOD however contributed to the poor outcome.[2–4]

Discussion

Mitochondrial fatty acid oxidative disorders are errors of metabolism inherited genetically in an autosomal recessive pattern. There are multiple types of FAOD's, some associated with pregnancy complications in the heterozygous mother, such as fetal growth restriction, severe preeclampsia spectrum, fatty liver of pregnancy, preterm delivery; these are most often noted in mothers carrying a fetus with a LC-FAOD. Newborns with the disorder typically present with abnormalities inclusive of: metabolic acidosis, hepatic failure, cardiac arrhythmias, neuropathy, retinopathy, hypoketotic hypoglycemia and cardiomyopathy. Definitive diagnosis resides with NBS for enzyme deficiency. Parents should be counseled that there is a 25% recurrence risk with future pregnancies. This case highlights the importance of a high suspicion of the disorder such that rapid multidisciplinary approach can be employed for the benefit of both patients and their parents.

Acknowledgments

None.

Conflicts of interest

None to disclose.

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