A Case of Metabolic Genetic Emergency

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Research Abstract Title

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Describe role of Submitting/Presenting Trainee in this project (limit 150 words):

This project was compiled as a case report as well as a professor rounds presentation.

Background, Objectives/Goal, Methods/Design, Results, Conclusions limited to 500 words

Background:
Newborn screening (NBS) is one of the largest public health initiatives to improve infant morbidity and mortality, and this has been shown to detect and lead to prompt treatment of uncommon but rapidly fatal conditions. While NBS has changed outcomes of diseases, it is still imperative that prompt and early recognition of abnormal results and medical interventions remain the mainstay of care.
Objective: To describe a case of inborn error of metabolism in a 4-day old asymptomatic infant, where early recognition and treatment affected outcome.
Design/Methods: This case is a 4-day old female, former 36-week gestational age infant who presented to her pediatrician's office for a routine well newborn exam.
Results: Birth history was unremarkable, and infant was discharged at 2 days of life. Prior to pediatrician visit, mother had no concerns. However, in the office, she was noted to be lethargic, hypothermic (T 35.4C) with weight down 8.3% from birthweight. Glucose was 48 mg/dL. While infant was in office, pediatrician was notified infant's NBS was positive for medium chain acetyl Co-A dehydrogenase deficiency (MCADD). Due to apparent metabolic crisis, she was admitted to the NICU where she required IV dextrose boluses and infusion of dextrose with a GIR of 15 mg/kg/min. Subsequently, she was allowed to feed at 200mL/kg/day. An acylcarnitine profile showed C8 level 15 times the normal value, which confirmed the diagnosis of MCADD. The infant was discharged at 2 weeks of life on a strict every 2.5 hour feeding regimen of breastmilk and is doing well.

This case report will discuss the following: how infants can be relatively asymptomatic at parent's glance

when suffering a metabolic crisis, the clinical and biochemical differences in MCADD, the history of NBS, burden on families of a metabolic diagnosis, and will evaluate differences in NBS in neonates admitted to the NICU versus well-baby nursery, as well as how to interpret the findings, and the timeline for follow up in different circumstances. This infant also highlights the imperativeness of NBS, and how NBS for relatively asymptomatic, uncommon, treatable conditions can drastically alter the course of life for a child and family. **Conclusion(s):** Notification of providers when an abnormal NBS is read is currently done via phone. However, in the 21st century of patient portals and apps, a digitization of newborn screen results could be utilized to improve patient outcomes. Though NBS has improved survival, physician understanding of signs and symptoms of metabolic crisis and emergent management is of paramount importance.