
A Newborn with Distended Abdomen

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CASE DESCRIPTION

A male neonate was born to a 35-year-old mother at an outside hospital at 39 1/7 weeks gestation via emergency cesarean section due to decreased fetal movements. The infant had respiratory distress secondary to meconium aspiration and was noted to have a large, distended abdomen at birth. Subsequent ultrasound confirmed hepatosplenomegaly with no evidence of liver mass or ascites. By report, alanine aminotransferase, aspartate aminotransferase, and alkaline phosphatase were initially within the reference intervals, but the patient had a prolonged INR (international normalized ratio) and hypoalbuminemia, supporting liver dysfunction. The infant was transferred to our institution for specialized care owing to hepatosplenomegaly, hypoglycemia, and coagulopathy of unknown etiology. On admission, the infant was additionally noted to be mildly pancytopenic, with increased lactate and ammonia. Initially, infectious etiologies were of primary concern given the history of meconium aspiration and the history of hepatosplenomegaly, which may be a presenting feature in patients with congenital infection. However, an extensive infectious disease workup was negative and included screens for hepatitis A, B, and C, HIV, herpes simplex virus, cytomegalovirus, Epstein–Barr virus, parvovirus, enterovirus, toxoplasmosis, and blood, urine, and stool cultures.

Hemophagocytic lymphohistiocytosis (HLH), although rare, was also a diagnostic consideration, as it can present with hepatosplenomegaly, coagulopathy, and pancytopenia, which were seen in this patient. However, the patient's pancytopenia resolved and the patient lacked additional features of HLH such as hypertriglyceridemia and hyperferritinemia. Given the patient's presentation of hepatosplenomegaly, hypoglycemia, and increased lactate and ammonia, an inborn error of metabolism was also part of the initial differential diagnosis. To screen broadly for metabolic disorders, plasma amino acids, urine organic acids, and plasma acylcarnitines were ordered stat while newborn screen results were pending. The clinical team was then called by the laboratory regarding the urine organic acid screen results and informed that although the overall urine organic acid profile was unremarkable, they noted a significant additional peak that identified as vanillylmandelic acid (VMA), and recommended quantification by a more specific method if clinically indicated. VMA and homovanillic acid (HVA) are catecholamine metabolites that are increased in patients with catecholamine-secreting tumors such as neuroblastoma, pheochromocytoma, and other tumors of neural crest origin. Given the lack of a mass on imaging studies of this patient, a malignant process was not high on the differential. Despite this, the team proceeded with the recommendation to specifically quantify HVA and VMA given the lack of a diagnosis in this patient at the time. Specific quantification demonstrated substantial increases in both urine VMA (430 mg/g creatinine, reference interval, 0–32.8 mg/g creatinine) and HVA (432.5 mg/g creatinine, reference interval, 0–17.6 mg/g creatinine).

QUESTIONS TO CONSIDER
<ul style="list-style-type: none">• What is in the differential diagnosis for a newborn with hepatomegaly?
<ul style="list-style-type: none">• Taken together with the patient's clinical presentation, what diagnosis do the HVA and VMA results suggest?
<ul style="list-style-type: none">• What additional tests or procedures are needed to establish the diagnosis in this patient?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the August 2016 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol62/issue8> and follow the link to the Clinical Case Study and Commentaries.

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