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Clinical Case Study

Elevated C3-Carnitine in a Healthy Premature Infant

Kimberly A Chapman,[1] Michael J. Bennett,[2] and Neal Sondheimer[3*]

Departments of [1]Genetics, [2]Pathology and Laboratory Medicine, and [3]Child Development, Rehabilitation, and Biochemical Genetics, The Children's Hospital of Philadelphia, Philadelphia, PA
*Address correspondence to this author at: The Children's Hospital of Philadelphia, 34th Street and Civic Center Blvd, Philadelphia, PA, 19104. Fax 215-590- 297; e-mail Sondheimer@email.chop.edu.

CASE DESCRIPTION

A 5-day-old male infant with an increased dried blood spot propionylcarnitine (C3-carnitine) value of 7.93 μmol/L (cutoff <6.79 μmol/L) was identified by the New Jersey state newborn screening program. C3-carnitine is used as a screening tool for methylmalonic and propionic acidemias, potentially fatal but treatable inborn errors of metabolism. The initial screen values provided a calculated C3:C2 carnitine ratio of 0.23 (cutoff <0.32, mean 0.074) and a C3:C16 ratio of 2.51 (cutoff <4.16, mean 0.96). The child was an inpatient at an outlying neonatal intensive care unit. He was born at 35 weeks estimated gestational age, required continuous positive airway pressure for a short time after birth, and transitioned quickly to room air. He was taking regular feedings with a cow's milk protein–based formula.

On day of life 6, the patient developed a mild acidosis (pH 7.24 on arterial blood gas testing). Because methylmalonic and propionic acidemia could not be excluded while confirmatory test results were pending, feedings were discontinued, intravenous hydration with glucose-containing fluids was initiated, and the infant was transferred to our institution. On arrival the child appeared well, was alert, and had normal growth parameters and no tachypnea. He had good tone and normal reflexes, and laboratory studies showed no acidosis. We allowed normal feedings and proceeded with the diagnostic evaluation.

Questions to Consider
• What is in the differential diagnosis for a newborn with elevated serum C3-carnitine?
• The largest group of defects associated with C3-carnitine elevation involves the downstream enzyme methylmalonyl-CoA mutase (MMM). What does MMM convert? What serves as a co-factor for MMM?
• What diagnostic laboratory tests can be used to help differentiate the causes of C3-carnitine elevations?

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Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the November 2008 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol54/issue11/ and follow the link to the Clinical Case Study and Commentaries.

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