

Vomiting and fasting: Risky in infancy

A nine-month-old healthy girl woke up from her nap with a low-grade fever and crankiness. She nibbled at dinnertime and started vomiting shortly after. She refused solids, did not tolerate liquids and continued vomiting repeatedly through the night. Because she was very pale, weak and progressively more somnolent, her parents decided to bring her to the emergency department in the early hours of the morning. On arrival, she had a generalized tonic-clonic convulsion. After initial resuscitation and stabilization, the emergentologist

found her physical examination to be normal except for an altered level of consciousness and mild hepatomegaly. Initial investigations revealed hypoglycemia and slightly elevated alanine aminotransferase and ammonia levels. An intravenous glucose drip was started and she progressively improved. The diagnosis of medium-chain acyl-coenzyme A dehydrogenase (MCAD) deficiency was confirmed with elevated serum acylcarnitine levels (C8) and the presence of an A985G mutation (homozygous).

LEARNING POINTS

- MCAD deficiency is the most common fatty acid oxidation disorder, with an incidence of approximately one in 10,000 to one in 20,000. The inheritance pattern is autosomal recessive.
- At initial presentation, the mortality risk is as high as 25%. Surviving infants often have residual neurological deficits and subsequent developmental delays.
- Prognosis is excellent with early diagnosis and treatment and, most importantly, strict avoidance of fasting. These factors are making a strong argument for universal newborn screening of this condition.
- Some Canadian provinces (British Columbia, Saskatchewan, Ontario, Nova Scotia and

Prince Edward Island) have implemented universal newborn screening with tandem mass spectrometry, which is able to detect as many as 40 metabolic disorders, including MCAD deficiency. The Canadian Paediatric Surveillance Program (CPSP) study is being conducted at an opportune time because it will allow comparisons between jurisdictions with and without universal newborn screening.

- The CPSP MCAD deficiency study was initiated in September 2005 and one case has been confirmed thus far. A parallel study is currently being done by the British Paediatric Surveillance Unit and will facilitate international comparisons.

The Canadian Paediatric Surveillance Program (CPSP) is a project of the Canadian Paediatric Society that undertakes the surveillance of rare diseases and conditions in children. For more information, visit our Web site at <www.cps.ca/cpsp> or <www.cps.ca/pcsp>.