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SHORT REPORT

Hyperlipidaemia due to carnitine palmitoyltransferase I deficiency

H. Worthington · S. E. Olpin · I. Blumenthal · A. A. M. Morris

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Summary We report a patient with carnitine palmitoyltransferase I (CPT I) deficiency, who presented with acute encephalopathy at 6 months of age. This was precipitated by an episode of gastroenteritis. No hypoglycaemia was documented, but there was hepatomegaly; blood tests revealed raised transaminases, a coagulopathy and severe hypertriglyceridaemia (48.8 mmol/L) and hypercholesterolaemia (9.5 mmol/L). The hyperlipidaemia resolved within 3 days of treatment and did not recur. At 2 years of age, the patient's liver function, growth and development are all normal. Hyperlipidaemia has been reported during acute illness in previous patients with CPT I deficiency but it is not a well-recognized feature; it should alert metabolic specialists to this potential diagnosis.

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H. Worthington · A. A. M. Morris (✉)
Willink Unit, Royal Manchester Children's Hospital, Manchester, UK
e-mail: Andrew.morris@cmmc.nhs.uk

S. E. Olpin
Chemical Pathology Department, Sheffield Children's Hospital, Sheffield, UK

I. Blumenthal
Paediatric Department, Royal Oldham Hospital, Oldham, UK

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SHORT REPORT

Late presentation of medium-chain acyl-CoA dehydrogenase deficiency

S. J. Mayell · L. Edwards · F. E. Reynolds · A. B. Chakrapani

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Summary Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is the commonest disorder of fatty acid metabolism, with a high incidence of morbidity and mortality at presentation. We report a 16 year old girl with first presentation of MCAD deficiency following an alcoholic binge and subsequent period of starvation. Presentation was as acute encephalopathy progressing to coma. Renal, cardiac and hepatic failures were managed with intensive supportive care including mechanical ventilation, inotropic support, blood products and renal replacement therapy. Diagnosis of MCAD deficiency was confirmed on day 6. The patient was discharged from hospital on day 20 with a mild proximal myopathy, which subsequently resolved. The diagnosis of MCAD deficiency requires a high index of suspicion at all ages. Precipitating factors in later life may include alcohol.

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S. J. Mayell · L. Edwards · F. E. Reynolds · A. B. Chakrapani (✉)
Department of Clinical Inherited Metabolic Disorders, Birmingham Children's Hospital, Steelhouse Lane, Birmingham B4 6NH, UK
e-mail: A.Chakrapani@bch.nhs.uk

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