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SHORT REPORT

Acute liver failure in pregnancy associated with maternal MCAD deficiency

L. Santos · A. Patterson · S. M. Moreea · C. M. Lippiatt · J. Walter · M. Henderson

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Summary In recent years the association between severe pregnancy complications and fetal fatty acid oxidation (FAO) disorders has been reported. However, there are few descriptions of a maternal FAO disorder leading to these complications. We describe acute liver failure associated with an undiagnosed maternal medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. The previously healthy proband presented at the 39th week with an itchy rash, palmar erythema and trace proteinuria; she was admitted onto a maternity ward. Acute fatty liver was suspected from the blood tests and a Caesarean section was performed, delivering a healthy boy. Cord blood samples were taken at delivery as part of an ongoing research project. The analysis of the cord blood sample showed a high concentration of octanoylcarnitine of 2.3 μmol/L (reference <0.1), suggesting a possible fatty acid oxidation disorder. However, subsequent acylcarnitine analyses of the baby's blood showed a normal pattern. The proband was further evaluated by urine organic acids and acylcarnitine profile. Elevated concentrations of hexanoylglycine in urine and octanoylcarnitine in blood spots were found, consistent with a diagnosis of MCAD deficiency. Mutation analyses confirmed that she was homozygous for c.985A>G (K329E). Even though these pregnancy complications are rare and it is not possible to affirm that the proband's acute liver failure was secondary to an undiagnosed MCAD deficiency, it seems likely.

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L. Santos (✉) · A. Patterson · C. M. Lippiatt · M. Henderson
Department of Clinical Biochemistry, St James University Hospital Leeds, Leeds, UK
e-mail: luciguimaraes@ntlworld.com

S. M. Moreea
Department of Gastroenterology, Bradford Royal Infirmary, Bradford, UK

J. Walter
Willink Biochemical Genetics Unit, Royal Manchester Children's Hospital, Manchester, UK

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