Transient neonatal multiple acyl-CoA dehydrogenase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Transient neonatal multiple acyl-CoA dehydrogenase deficiency. ORPHA:329942

Transient neonatal multiple acyl-CoA dehydrogenase deficiency describes a very rare condition where a maternal riboflavin deficiency causes an infant to present with manifestations similar to those seen in multiple acyl-CoA dehydrogenase (MAD) deficiency (see this term) such as poor suck, metabolic acidosis and hypoglycemia, but that resolves completely with oral riboflavin. In the one patient described haploinsufficiency of the human riboflavin transporter (hRFT 1) was described in the mother.