INHERITED METABOLIC DISORDERS (IMD)
Supporting information

This guideline has been produced with reference to the following:

British Inherited Metabolic Disorders Group. Guidelines: Emergency Protocols- Children. BIMDG

http://www.bimdg.org.uk/site/guidelines.asp

Gospe SM. Pyridoxine-Dependent Epilepsy in GeneReviews. 2001 (updated 2021)

https://www.ncbi.nlm.nih.gov/books/NBK1486/

Metabolic Evaluation of Epilepsy: A Diagnostic Algorithm With Focus on Treatable Conditions
van Karnebeek CDM,Sayson B, Lee JJY et al. Front Neurol. 2018; 9: 1016

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6286965/

Al-Baradie RS, Chaudhary MW. Diagnosis and management of cerebral folate deficiency. A form of folinic acid-responsive seizures. Neurosciences (Riyadh). 2014;19:312-6

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4727671/

Zschocke J & Hoffmann GF. Vademecum Metabolicum: Diagnosis and Treatment of Inborn Errors of Metabolism. 2011. Schattauer Gmbh

http://www.vademetab.org/

Stockler S, Plecko B, Gospe SM et al. Pyridoxine dependent epilepsy and antiquitin deficiency: clinical and molecular characteristics and recommendations for diagnosis, treatment and follow-up. Mol Genet Metab. 2011;104:48-60

Leonard JV, Morris AA. Diagnosis and early management of inborn errors of metabolism presenting around the time of birth. Acta Paediatr. 2006;95:6-14

Ficicioglu C, Bearden D. Isolated neonatal seizures: when to suspect inborn errors of metabolism. Pediatr Neurol. 2011;45:283-91

Saudubray JM, Nassogne MC, de Lonlay P et al. Clinical approach to inherited metabolic disorders in neonates: an overview. Semin Neonatol. 2002;7:3-15

Clayton PT. Inborn errors presenting with liver dysfunction. Semin Neonatol. 2002;7:49-63

Burton BK. Inborn errors of metabolism in infancy: a guide to diagnosis. Pediatrics. 1998;102:E69.

https://pediatrics.aappublications.org/content/102/6/e69.long

Hoffmann GF, Surtees RA, Wevers RA. Cerebrospinal fluid investigations for neurometabolic disorders. Neuropediatrics. 1998;29:59-71

Treatment with L-carnitine is appropriate in the management of neonatal hyper ammonaemia, organic acidaemia, fatty acid oxidation disorders and lactic acidosis?
A Cochrane systematic review (Nasser, 2012) was unable to identify any randomised trials in this area and concluded that, “in the absence of any high level evidence, clinicians should base their decisions on clinical experience and in conjunction with preferences of the individual where appropriate. This does not mean that carnitine is ineffective or should not be used in any inborn error of metabolism. However, given the lack of evidence both on the effectiveness and safety of carnitine and on the necessary dose and frequency to be prescribed, the current prescribing practice should continue to be observed and monitored with care until further evidence is available.”

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Nasser M, Javaheri H, Fedorowicz Z, et al. Carnitine supplementation for inborn errors of metabolism. Cochrane Database of Systematic Reviews 2012,Art. No.: CD006659
http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD006659.pub3/full

Evidence Level: I (For "no evidence")

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