Abstracts

The poster incorporated a check list on how to assess a neonate for signs of deterioration under the headings: Breathing, Colour, Temperature, Activity and Feeding. This was then taught to the nurses using clinical videos and bedside teaching in the neonatal unit. The nurses were asked to complete a test assessing their knowledge before and after the teaching session. The mean scores were compared using the student t-test to determine if there was any statistically significant difference.

Result The data shows that there was a statistically significant improvement in the test scores after training in both hospitals. In Hospital A, the average test score before training was 61.3% but improved to 81.3% after training (p=0.007). In Hospital B the average test score before training was 83.3% and then 97.8% after training (p= 0.016).

Conclusion The training programme was effective in improving nursing knowledge in the recognition of the sick and deteriorating neonate as well as escalation of care.

G469 DEVELOPMENTAL DELAY IN A YOUNG INFANT WITH NON-CLASSICAL COMBINED MALONIC AND METHYL MALONIC ACIDURIA (CMAMMA) CAUSED BY HOMOZYGOUS MISSENSE MUTATION IN ACSF3 GENE

1MBKC Dayasiri, 1E Goh, 1S Kodagali, 2J Baruteau, 1G Anand. 1Department of Paediatrics, Oxford University Hospital, Oxford, UK; 2Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital, Great Ormond Street Hospital, UK

10.1136/archdischild-2020-rcpch.402

Introduction AcylCoA synthetase family member 3 (ACSF3) activates malonylCoA and methylmalonylCoA into their respective thioesters. ACSF3 deficiency causes non-classical CMAMMA, a rare inborn error of metabolism characterised by presence of methyl malonic acid in high concentrations than malonic acid in urine. The reversal is seen with classic CMAMMA caused by malonylCoA decarboxylase deficiency (MCD).

Case Report 5 and a half month old male infant second born to consanguineous South Asian parents presented with severe failure to thrive and recurrent vomiting. His older sibling who had failure to thrive and neuro-developmental delay died at seven months without a genetic diagnosis.

Initial blood tests revealed metabolic acidosis, pancytopenia and coagulopathy. Neuroimaging was unremarkable. Subsequent evaluation revealed normal levels of methionine, homocysteine and red cell folate. Significant methylmalonic aciduria with mild malonic aciduria without evidence of other abnormal metabolites (propionyl-CoA metabolites: hydroxypropionate, and methylcitrate or tiglylglycine) in urine suggested the diagnosis of non-classical CMAMMA, confirmed by homozygous missense variants in ACSF3 gene revealed by trio-exome sequencing. Neurodevelopmental assessment at eight months revealed global developmental delay with general hypotonia; gross motor (4–6 months); fine motor (4–6 months); speech (5–6 months) and social (under 6 months), and without any regression. Carnitine was supplemented to avoid secondary depletion caused by the excretion of MMA. Parents were advised to avoid prolonged fasting and to provide emergency regimen (powdered carbohydrate drink mix) in the event of acute deterioration.

Conclusion This report describes an unusual paediatric presentation of non-classic CMAMMA. Urine organic acids allows identification of increased MA and MMA excretion and highly suggestive of the diagnosis, thus avoiding additional investigations. Determination of urinary MMA/MA ratio can help differentiating between classical and non-classical forms.