MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCADD) – EARLY MANAGEMENT OF BABIES WITH FAMILY HISTORY

See British Inherited Metabolic Disorders Group (BIMDG) Guideline: **Management of newborn babies who might potentially have MCAD deficiency** (https://bimdg.org.uk/)

DEFINITION

- Inherited disorder of fat metabolism
- With regular intake of food, individuals can lead a normal healthy life but prolonged fasting or illness with vomiting can lead to encephalopathy, coma or sudden death
- Hypoglycaemia is a late sign
- Babies with a family history of MCADD require a special feeding regimen and observation from birth
- Families of newborn babies with family history of MCADD should have already received correspondence from specialist metabolic centre in advance of birth, regarding additional feeding support/investigations required

BACKGROUND

- When mother admitted in labour, inform neonatal team
- When baby aged 24–48 hr, carry out following tests:
- bloodspot acylcarnitines
- urine organic acids
- DNA mutation analysis [in most cases, specific genotype will be known for the index case – write this (and details of index case) on molecular genetics form]
- Discuss testing with metabolic laboratory at Birmingham Children's Hospital and mark request 'family history of MCADD'.
- If baby on glucose IV at time of sampling, repeat acylcarnitine sample **must** be taken after 24 hr of full feeds (to avoid false negative)
- Ensure full routine newborn bloodspot screening completed on day 5

MANAGEMENT

- Any at risk baby **must** receive sufficient milk intake to avoid potential for decompensation while awaiting results
- Specific feeding advice depends on whether baby is bottle/breastfed
- See **Management of newborn babies who might potentially have MCAD deficiency** (https://bimdg.org.uk/) for specific feeding guidance

PROBLEMS

- **If baby becomes drowsy or unwell in any way, admit to NNU urgently**
- Follow **Management of newborn babies who might potentially have MCAD deficiency** (https://bimdg.org.uk/)
- Seek urgent advice from specialist metabolic centre

DISCHARGE

- Assess baby's feeding; ensure reliably feeding well before considering discharge – continue special feeding regimen until results available
- If baby feeding well, risk of neonatal decompensation is low after 72 hr
- Baby can be safely discharged before this, even if results are not known, provided feeding well
- Give parents clear safety-netting instructions – return to hospital if feeding is poor

LOCAL CONTACT

- For specialist advice, consult on-call metabolic registrar/consultant via Birmingham Children's Hospital switchboard (0121 333 9999)

FURTHER INFORMATION

BIMDG website: http://www.bimdg.org.uk/guidelines.asp