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

Based on British Inherited Metabolic Disease Group Protocol updated Nov 2018

DEFINITION

- A rare autosomal recessive inherited metabolic disease where the body cannot metabolise fat properly
- 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
- Affects 1:10,000 babies in UK. 1:80 healthy people are carriers
- Bloodspot screening at day 5 includes MCADD (see **Bloodspot screening** guideline)
- Newborn babies with MCADD are especially vulnerable in first few days of life before breast milk supply and regular feeding pattern established
- Babies with a family history of MCADD may require a special feeding regimen and observation from birth. If one parent is affected, the baby can be treated a normal. Only if a sibling is affected (as we then assume that both parents are carriers) does the baby require a feeding plan.

SYMPTOMS

- Often non-specific
  - hypothermia
  - jitteriness
  - irritability
  - drowsiness
  - reluctance to feed
  - lethargy
  - rapid breathing
  - seizures
  - coma
  - sudden death
- Hypoglycaemia occurs late

DIAGNOSIS

- When mother admitted in labour, inform neonatal team
- Test baby aged 24–48 hr
  - bloodspot acylcarnitines
  - urine organic acids
  - DNA mutation analysis (in most cases, genotype will be known for the index case)
- Discuss testing with metabolic laboratory at Birmingham Children's Hospital and mark request 'family history of MCADD'
- Continue special feeding regimen until results available

MANAGEMENT

- High index of suspicion antenatally
- Refer those with family history of MCADD for genetic counselling antenatally
- Advise parents baby may require specialist feeding regimen from birth and rapid testing at aged 24–48 hr
- Institute specialist feeding regimen from birth and ensure regular milk intake
- Complete bloodspot screening as normal on day 5

- If baby not meeting target volumes start nasogastric tube feeds
- If enteral feeds not tolerated, commence IV fluid – glucose 10%, sodium chloride 0.18%
- Routine monitoring of blood glucose not necessary
- Bottle fed babies
  - term baby: 4-hrly feeds
  - preterm baby: 3-hrly feeds
  - fluid intake should be 60 mL/kg/day on day 1, increasing to 150 mL/kg/day by day 7
- Breastfed babies
  - at particular risk in first 72 hr
  - should breastfeed for ≥10 min, 8 times/day
  - observe feeding to check baby latched on well and has a slow rhythmic suck (i.e. good technique)
  - give all breastfed babies formula top-ups until good maternal milk supply established
    - day 1: 25 mL/kg
    - day 2: 40 mL/kg
    - day 3: 60 mL/kg
  - if baby not taking adequate oral feeds, start nasogastric tube feeding

PROBLEMS

- If baby drowsy or unwell in any way, admit to NNU urgently
  - give 2 mL/kg glucose 10% as IV bolus, then commence infusion of glucose 10% at 100 mL/kg/day
  - change to glucose 10% with added sodium chloride from day 3
  - if no oral intake increase IV infusion to 150 mL/kg/day over 3 days
  - monitor blood glucose and electrolytes, but base treatment on clinical state as hypoglycaemia occurs late
- Seek advice from specialist metabolic centre

DISCHARGE

- Assess baby's feeding before considering discharge
- Give parents clear instructions to return to hospital if feeding is poor
- 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 baby's feeding is secure

LOCAL CONTACT

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

FURTHER INFORMATION

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