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

Based on British Inherited Metabolic Disease Group Protocol

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 require a special feeding regimen and observation from birth

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 will require specialist feeding regimen from birth and rapid testing at aged 24–48 hr
- Institute specialist feeding regimen from birth
- Ensure regular milk intake
- term baby: 4-hrly feeds
- preterm baby: 3-hrly feeds

- Breastfed babies are at particular risk in first 72 hr. Give 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
- If enteral feeds not tolerated, commence IV fluid – glucose 10%, sodium chloride 0.18%
- Complete bloodspot screening as normal on day 5

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
- 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

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