Hyperammonaemia

Refer EARLY to KIDS NTS for advice - 0300 200 1100

Is this a TIME-CRITICAL transfer? - HYPERAMMONAEMIA needs URGENT discussion with KIDS NTS + BCH metabolic consultant

History: antenatal diagnosis; consanguinity; family history; SIDS or multiple miscarriages; increased foetal movements (seizures); a period of normality prior to decompensation is common.
Clinical Features: hypotonia; seizures; encephalopathy; lethargy; poor feeding; hypoglycaemia; vomiting; diarrhoea; dehydration; hepatomegaly; jaundice; cardiac failure .
Biochemical Features: metabolic acidosis; respiratory alkalosis; hypoglycaemia; hyperlactatemia; hyperammonemia.
Differentials: important to consider other causes of **neonatal collapse**. (i.e sepsis, congenital heart disease)



Airway/Breathing

Support airway; administer O2 to maintain sats > 92%; monitor respiratory rate and SpO2; Intubate and ventilate as required (i.e low GCS and lack of airway reflexes).



Circulation

Monitor: HR, BP, UO, perfusion and pulses; obtain 2 points of venous access; use 0.9% saline boluses to support circulation; consider vasoactive agents after 40 ml/kg.



Disability

Treat Hypoglycaemia - 2ml/kg 10% dextrose; treat seizures as per APLS algorithm; assess pupil size; consider cranial imaging - i.e CrUSS if haemofiltration required.

Hyperammonaemia: Can result in **CEREBRAL OEDEMA, SEVERE NEUROLOGICAL IMPAIRMENT and DEATH**.

By definition - **Neonates** > 100umol/l (and > 200umol/l is significantly abnormal)
Infant/older child > 50umol/l (and > 100umol/l is significantly abnormal)

Lab testing - **URGENT**. Free flowing venous or arterial sample (NOT capillary); needs delivery by person on ICE to the lab, with immediate processing by lab within 15 minutes. Please liase with lab before and after sample taken.

Dependent on the ammonia level, medical management and/or haemofiltration may be required.

Specific Management:

- Stop oral feeds.
- Refer to BIMDG Emergency Guidelines at https://bimdg.org.uk/ for “undiagnosed problems” or "emergency management of specific conditions where known".
- IV maintenance fluids as per https://bimdg.org.uk/
- Start emergency metabolic drugs as per https://bimdg.org.uk/ – in discussion with KIDS/Metabolic Consultant
- Arrange transfer to specialist centre as soon as possible.
- Frequent blood gas monitoring including: blood glucose & lactate.
- Send urgent venous or arterial plasma ammonia as per the above instructions.
- Check FBC, CRP, UE, CK, LFTs & Coag.
- Send Lithium Heparin blood sample & collect urine for metabolic screen (amino acids, acylcarnitines & organic acids) – these samples can accompany the child to the specialist centre.
- If possible-additional EDTA (purple x2) samples can be taken to save for genetics if required.
- Take Cross Match – may require Haemofiltration.

Pharmacy and KIDS NTS metabolic drugs bag:
- If specialist drugs required, local pharmacist should be called and can liase with BCH pharmacist via (0121 333 9999).
- KIDS NTS stock some emergency metabolic scavenger medications, and may be able to help provide these to local teams. However, do not wait for specialist drugs to be available if urgent transfer is required for specialist treatment such as haemofiltration.

Author - KK/RN - Sept 26, review Sept 30