NHS CATS
Children's Acute Transport Service

Clinical Guideline

Metabolic Referrals

Document Control Information

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Establish if a metabolic condition is known and to which centre. Known metabolic conditions should be discussed with the duty GOSH (or base centre) metabolic consultant. If a new metabolic condition is suspected, early discussion with the metabolic on-call team is advisable, especially for hyperammonaemia.

Assessment

Presentation

- Encephalopathy, seizures and apnoea
- Metabolic acidosis but *NB* most metabolic acidosis is due to poor tissue perfusion (sepsis/low cardiac output) rather than metabolic disease.
- Hypoglycaemia
- Hypocapnia with tachypnoea
- Cardiac failure/cardiomyopathy
- Liver dysfunction
- Dysmorphism
- Hydrops
- Hyperammonaemia

History and clinical examination

Ask about:

- Involvement of a metabolic team
- Family history of metabolic disorder
- Previous pregnancy losses
- Consanguinity
- Any problems during the pregnancy
- Poor feeding, growth, or development

Examination should focus on cardiorespiratory and neurological status.

Initial screening tests for metabolic disease

- Odour of baby and urine
- Blood glucose
- Venous sample for plasma ammonia (trend data are important)
- Acid-base status
- Anion gap ([Na+ + K+] - [Cl- + HCO3-] (normal <12 mmol)
- Lactate (trend data are important)
- Liver function tests

- Creatine kinase
- Urinary/plasma ketones (point of care urinalysis or capillary ketones)

Initial management

If a metabolic disease is suspected, the case should be discussed with the duty GOSH consultant in metabolic medicine.

Supportive therapy

- Profound encephalopathy, intractable seizures or apnoea – intubate and provide mechanical ventilation. Consider pre-intubation CO2.
- Circulatory failure - intravascular volume expansion (5-10mls/kg aliquots with reassessment between) ± vasoactive therapy.
- Stop oral feeds and give 10% glucose ± electrolytes.
- Give glucose at an infusion rate of at least 7mg/kg/min to promote anabolic state.
- If hyperglycaemic, may require insulin.
- If hypoglycaemic, give glucose infusion to maintain euglycaemia (may need to use a glucose concentration >10%, ideally through a central venous catheter).
- Seizures – treat hypoglycaemia, commence APLS protocol anticonvulsants if necessary, consider hyperammonaemia.
- Metabolic acidosis – consider correcting pH using sodium bicarbonate (discuss with metabolic team).
- Hyperammonaemia - sodium benzoate (IV), sodium phenylbutyrate (IV), L-arginine (IV), L-carnitine (IV), should be brought on transport. Refer to IV infusion loading and maintenance doses below. In cases of hyperammonaemia with unknown cause, loading doses of all the above should be administered followed by maintenance infusion. Early discussion with the metabolic team is advised. Additionally, consider taking carglumic acid (enteral) if available in discussion with the metabolic team.

Indications for intubation

- Depressed level of consciousness
- Intractable seizures
- Apnoea
- Severe circulatory failure
- Need for haemofiltration

Management following intubation

- Sedate and paralyse
- Ensure ETT well secured
- Capnography should be used to confirm tube placement and for ongoing monitoring of end-tidal CO_2
- Confirm correct ETT position with CXR
- Attain adequate venous access, consider arterial access, in neonates consider UAC and UVC
- Give sodium bicarbonate if acidosis intractable and CO_2 allows

Samples to bring with patient (if requested by metabolic team)

- Lithium heparin (2ml) for plasma amino acids
- Urine in standard urine container (pre-treatment sample very helpful for diagnosis, will be sent for urine organic acids)
- Blood spot on Guthrie card for carnitine profile if possible

Transport considerations

- Prepare fluid boluses
- Consider preparing infusion of adrenaline ready to commence immediately if necessary
- Administer appropriate metabolic drugs
- Be prepared to treat seizures and hypoglycaemia

Metabolic Drugs: *[Refer to details in BIMDG Guidelines[1]]*

- Sodium phenylbutyrate, sodium benzoate, arginine, levocarnitine and glucose may all be piggybacked / given via the same y-connector.
- Monitor Na levels.
- Discuss with metabolic team regarding the need for loading dose (these may be omitted if patient already receiving the drug) and ongoing infusion.

Sodium Benzoate:

(2g in 10ml). Load: 250mg/kg over 90 mins then 10-20 mg/kg/hr infusion. Dilute 2g in 40ml with 10% glucose (max concentration).

Sodium Phenylbutyrate:

(2g in 10ml). Load: 250mg/kg over 90 mins then 10-20 mg/kg/hr infusion. Dilute 2g in 40ml with 10% glucose (max concentration).

Levocarnitine:

(1g in 5ml). Load: 100mg/kg over 30 mins, then 4mg/kg/hr infusion. Dilute 1000mg in 50ml with 10% glucose.

Arginine:

(5g in 10ml). Load: 150mg/kg over 90 mins, then 12.5mg/kg/hr infusion. Dilute 2.5g in 50ml with 10% glucose, max concentration 50mg/ml. (Metabolic team may suggest higher doses in certain known conditions after discussion with metabolic consultant).

References

1. BIMDG (2024). British Inherited Metabolic Disease Group emergency guidelines: https://bimdg.org.uk/emergency-guides/