BIMDG

British Inherited Metabolic Disease Group

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This protocol has 4 pages

MANAGEMENT OF A BABY AT RISK OF HYPERAMMONAEMIA (UNDIAGNOSED) AT BIRTH.

- **This protocol is only intended to be used for <u>undiagnosed</u> hyperammonaemia.**
- **For those known to be at risk of a urea cycle disorder or organic acidaemia please refer to the appropriate protocol.**

1. The differential diagnosis of hyperammonaemia in the newborn period is wide. For details see the separate protocol on the BIMDG website. This protocol outlines the management. A careful history is essential and should be reviewed by a specialist. In particular when did the previous child become ill, in the neonatal period or later? Is the likely diagnosis known? Work out the quickest way to establish the diagnosis. Seek specialist help if necessary.

2. If a previous sibling became ill shortly after birth, discuss management with specialist.
   - Consider transferring the mother before birth to a centre with all facilities for managing an affected baby.
   - Consider delivering the baby by caesarian section as this minimizes the metabolic stress of birth and the timing of the delivery is known.

3. In the third trimester obtain supplies of the medicines used for the treatment of organic acidaemias and urea cycle disorders – enteral and intravenous carnitine, sodium benzoate, sodium phenylbutyrate and arginine. See if your pharmacy is willing to obtain a supply of N-carbamylglutamate (1g, enteral only), as this can correct hyperammonaemia in organic acidaemias.

4. Inform the clinical biochemistry laboratory about the impending birth, as it is essential that results are available quickly.

At this stage the management will depend on the illness in the previous sibling.

If the previous sibling became ill in the neonatal period: **Go to section A on page 2**
If the previous sibling became ill <u>after</u> the neonatal period: **Go to section B on page 4**

SECTION A

IF THE PREVIOUS SIBLING BECAME ILL IN THE NEONATAL PERIOD:

A1. Transfer to the neonatal unit immediately after birth and start an intravenous infusion of 10% glucose at 4 ml/kg/hr (6.6 mg/kg/min) as soon as possible , preferably within 30 minutes of birth. This is to prevent the normal fall in blood glucose that initiates catabolic pathways in the baby.

A2. If the baby remains well, at 4 hours offer a milk feed (breast or infant formula) but continue the intravenous infusion.

A3. At 6 hours of age measure plasma ammonia and then give carnitine 50 mg/kg, sodium benzoate 50 mg/kg and l-arginine solution 100mg/kg orally and continue the same dose 6 hourly thereafter until the diagnosis is known or advised to change.

If the plasma ammonia remains normal (< 80 μmol/l)

- **R**epeat in 6 hours and provided it remains normal, monitor at 12 hourly intervals.
- Continue to offer milk feeds approximately 4 hourly.
- Stop the intravenous infusion if the baby is well and the tests are normal at 24 hours of age.

If the values show minor abnormalities (e.g. ammonia 80-150 μmol/l)

- Repeat in 4 hours and if they persist, monitor at 6 hourly intervals.
- Stop the milk feeds and instead give feeds as 10% soluble glucose polymer. (Click here for instructions on making 10% Glucose Polymer Solution)
- Continue the intravenous dextrose infusion.

If any of the results are clearly abnormal (for example plasma ammonia > 150 μmol/l) and/or if the baby becomes clinically unwell repeat the tests immediately and seek advice from the metabolic centre.

A4. At approximately 12- 24 hours of age measure plasma ammonia, plasma aminoacids (quantitative) and blood spot acylcarnitines and urine organic acids urgently.

Note: The treatment proposed may mask the biochemical changes of disease. Careful follow-up is essential.

A5. IF UNWELL

- Contact the specialist centre.
- Try to get the results of the investigations as soon as possible.

Depending on the clinical state and the results consider the following:

- Stopping all feeds
- Monitoring plasma ammonia concentrations at least 4 hourly.
- Increasing the intravenous glucose infusion
- Giving medicines that may include carnitine, sodium benzoate, sodium phenylbutyrate and arginine intravenously and N-carbamylglutamate orally.
    - For more information about medicines click here.
    - For drug dose calculator click here.

- Transferring to a hospital with facilities for specialist care including haemodialysis/filtration.

A6. If mother wishes to breast feed, she should express as she should be able to breast feed her baby, even if affected, once the metabolic state is stable.

SECTION B

IF THE PREVIOUS SIBLING BECAME ILL AFTER THE NEONATAL PERIOD:

B1. If the birth is complicated (birth asphyxia, etc) start a glucose infusion as soon as possible after birth. This is to prevent the normal fall in blood glucose that initiates catabolic pathways in the baby. Admission to SCBU for assessment is advisable. Complications not only may be responsible for symptoms that mimic those of hyperammonaemia but they also elevate plasma ammonia concentrations.

B2. If all proceeds normally, start milk feeds (breast or infant formula).

B3. At 24-48 hours of age measure blood gases, plasma ammonia, aminoacids (quantitative) blood spot carnitine profile and urine organic acids and ketones. Continue with regular feeds.

B4. If any of the results are abnormal and/or the child is unwell at any time (refusing to feed, tachypnoeic, drowsy, floppy, vomiting, etc), **Repeat the tests at once at once and follow the instructions on A5.**

If there are minor abnormalities (for example plasma ammonia 80 -150 μmol/l) and the baby appears well

- Repeat at 12 hourly intervals.
- Try to get results of the investigations as soon as possible.
- Change feeds to 10% soluble glucose polymer (Click here for instructions on making 10% Glucose Polymer Solution)

If the plasma ammonia remains less than 80 μmol/l at 48 hours

- Continue milk feeds
  Discuss follow up at specialist centre.

B5. If mother wishes to breast feed, she should express as she should be able to breast feed her baby, even if affected, once the metabolic state is stable.

B6. Get the results.but note that the treatment proposed may mask the biochemical changes of disease. Careful follow-up is essential.

More information about the individual disorders can be found on the BIMDG Website

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