BIMDG
British Inherited Metabolic Disease Group

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This protocol has 2 pages

MANAGEMENT OF A BABY AT RISK OF MAPLE SYRUP URINE DISEASE AT BIRTH (MSUD)

1. The pregnancy should be allowed to proceed normally.

2. Management before birth.

i) If a previous sibling became ill shortly after birth, (Usually the illness has a sub-acute onset with illness presenting after a few days).

ii) Work out the quickest way to establish the diagnosis which will usually be measuring the plasma branched chain amino acid concentrations (see A4 for more details). Inform the clinical biochemistry laboratory about the impending birth, as it is essential that results are available quickly.

iii) Seek specialist advice if there are any concerns or uncertainties.

3. At birth 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
If the previous sibling became ill after the neonatal period: Go to section B on page 2

Section A

Note: Complications, such as asphyxia, not only may be responsible for symptoms that mimic those of MSUD and they also may be responsible for an increase in branched chain amino acids concentrations

A1. Feeding: The baby should be fed as guided by a specialist metabolic dietitian with a mixture of:

- 50% BCAA-free feed
- 50% breastmilk or standard infant formula
- 75mg isoleucine daily *(to facilitate earlier detection of raised alloisoleucine in affected babies)*

A2. At 12 - 24 hours of age measure plasma aminoacids (quantitative).

A3. Management
⇨If the results are within normal limits, start normal milk feeds and repeat plasma aminoacids 12-24 hours later.

⇨If the values are borderline, continue with 50% BCAA-free feed, 50% breastmilk or standard infant formula and 75mg isoleucine daily as guided by a specialist metabolic dietitian (additional valine and isoleucine supplements may be required) and repeat plasma amino acids in 12- 24 hours.

⇨If the results are clearly abnormal and/or if the baby becomes clinically unwell repeat the plasma amino acids immediately and seek advice from the metabolic centre.

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

Section B: If the previous sibling became ill <u>after</u> the neonatal period:

B1. If all proceeds normally, start milk feeds (breast or infant formula).

B2. At 24-48 hours of age measure plasma aminoacids (quantitative). Continue with regular feeds.

B3. If any of the results are clearly abnormal and/or the child is unwell at any time (refusing to feed, tachypnoeic, drowsy, floppy, vomiting, etc), repeat the tests at once and follow the instructions in A3. For more information about the feeds seek help of specialist dietitian.

If there are minor abnormalities of plasma branched chain aminoacid concentrations and the baby appears well repeat them. Try to get results of the investigations quickly.

B4. 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.

B5. Check plasma branched chain amino acid concentrations once more before the baby is allowed home.

Reference: DiGeorge AM, Rezvani I, Garibaldi LR, Scwartz M (1982) Prospective Study of Maple Syrup Urine disease for the first four days of life. New Engl J Med 307;1492- 1495

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