MSUD clinical management guidelines*

Presumptive positive MSUD**

Lab to contact specialist / designated team as per local protocol. The following information MUST be provided to the clinical team by the screening laboratory:

(a) Hospital of Birth
(b) Parent telephone number
(c) Telephone number of the midwife (office and ward for out of hours contact)

ON THE SAME DAY

1. Specialist team to CONTACT FAMILY to arrange urgent hospital admission **If family CANNOT BE CONTACTED**, see footnote ♦♦♦
2. Specialist team to instruct family to go to appropriate hospital with 24 hr paediatric cover.
3. Specialist team to liaise with the local hospital (on call Paediatric Consultant, or registrar or equivalent grade if unable to contact)
   a. Fax/email information to the hospital for clinicians and parents, BIMDG MSUD guidelines, 'MSUD is suspected' leaflet, contact numbers for the MSUD specialist team
   b. Clinical assessment and admission to hospital **regardless of clinical status**
      Obtain blood gases, U&E, LFT, FBC, cultures, urine ketones dipstick. Site IV cannula
   c. Hospital to liaise with specialist centre regarding clinical status
   d. Commence clinical management:
      i. IV 10% dextrose/0.45% saline +added potassium infusion
      ii. Transfer to specialist centre. If GCS <8, intubate, ventilate and organize Paediatric intensive care retrieval
      iii. If transfer not possible same day, obtain diagnostic samples*** and courier urgently to specialist centre laboratory. Specialist team to liaise with laboratory to expect samples from admitting hospital
      iv. If transfer not possible same day, specialist team to organise supplies of MSUD Anamix Infant formula, Isoleucine and Valine sachets and feeding plan****
   e. Continue liaison between specialist and local hospital until transferred
4. Specialist team to inform GP (as soon as practicable), send MSUD GP letter via fax / email
5. Specialist team to inform maternity services and health visiting services

FIRST REVIEW within 24 hours of screening result
If not in specialist centre, speak directly via telephone or other communication
Review available test results
(If at DGH, do not discharge until agreed by specialist team)

Alloisoleucine Present?

Original NBS card Alloisoleucine Present?

MSUD UNLIKELY
Presumptive false positive
Investigate and treat
Exclude liver disease including galactosaemia

Possible intermittent MSUD
Send for genetics/enzymology
Manage as MSUD until result known

MSUD CONFIRMED
Clinical management via specialist team
Arrange sibling screening*****

See **MSUD initial clinical referral guidelines and standards** for further details

** See **MSUD screening protocol** for details

*** See **MSUD diagnostic protocol** for confirmatory test details

**** See **MSUD dietetic management pathway** on BIMDG website - www.bimdg.org.uk/site/guidelines-enbs.asp

***** See **MSUD sibling protocol** for details

♦♦♦ If family cannot be contacted directly, consider the following options:

- Ascertain if baby in hospital of birth/hospital closest to home (these may not be the same) & contact appropriate paediatric consultant /registrar.
- Contact GP, midwife or community midwifery team (via labour ward where baby born if necessary), for information and ask if home visit possible; alternatively, family to be contacted with instructions to take baby immediately to local A&E for evaluation
- Leave message on phone and ask to call back. Phone again after 1 & 2 hours if no contact
- If these measures fail, contact police via 999 to establish whereabouts of the family