**MSUD clinical management guidelines**

**Presumptive positive MSUD**

Lab notifies Clinical Liaison Service (CLS) as per local protocol on the day of the result.

**ON THE SAME DAY**

1. CLS contact specialist team as per local protocol
2. Specialist team to CONTACT FAMILY to arrange urgent hospital admission
3. Specialist team to instruct family to go to appropriate hospital with 24 hr paediatric cover. Offer to arrange an ambulance
4. 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
5. Specialist team to inform GP (as soon as practicable), send MSUD GP letter via fax / email
6. 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)

* 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

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**Alloisoleucine Present?**

- **Yes**

**Original NBS card Alloisoleucine Present?**

- **Yes**

- **No**

**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*****