

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)

Alloisoleucine
Present?

Yes

No

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*****

* 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