Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Maple Syrup Urine Disease (MSUD) Clinical Referral Guidelines for Newborn Bloodspot Screening Wales

Based on the NHS Newborn Blood Spot Screening programme's: A laboratory guide to newborn blood spot screening for inherited metabolic diseases

[https://www.gov.uk/government/publications/newborn-blood-spot-screening-laboratory-guide-for-imds]

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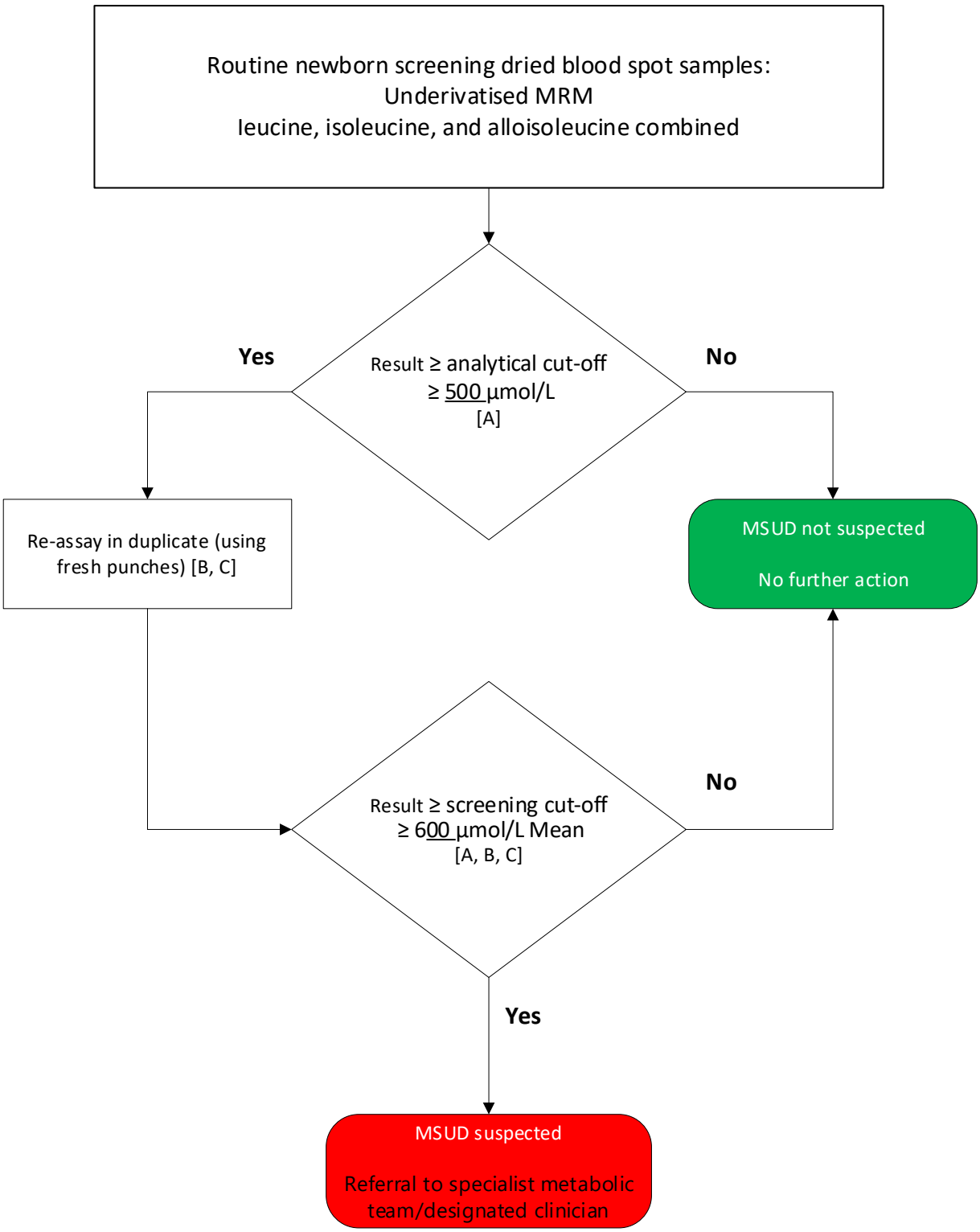
MSUD newborn screening protocol



Stage of Process	Guidelines
Defining an MSUD suspected screening result:	If a sample from a baby is found to have a leucine, isoleucine and alloisoleucine combined result equal to or greater than an analytical cut off of 500 µmol/L, repeat tests should be performed in duplicate on the original bloodspot card. If the mean of triplicate results is equal to or greater than 600 µmol/L, this is a presumptive positive screening result.
Referral of babies with a positive ('MSUD suspected') screening result:	**Betsi Cadwaladr UHB referrals will be to Manchester specialist metabolic team. For all other health boards in Wales the referrals will be sent to Birmingham Children's Hospital specialist metabolic team.** Wales Newborn Screening Laboratory notifies the local designated paediatrician AND the specialist metabolic team on the day of the result and also makes contact with the local laboratory to inform them.This includes referral on Saturdays where a result can be reported from samples analysed overnight on Friday. The clinical referral must be both verbally (by telephone) and by an email using a standard letter. The following information must also be emailed to the local designated paediatrician along with the standard letter. • 'MSUD is suspected' leaflet • 'MSUD GP letter • 'MSUD A&E letter If a disorder other than MSUD is suspected referral must be made to the specialist metabolic team.

First family contact	The Local designated paediatrician to contact the specialist metabolic team (it is important that the local designated paediatrician is guided by expert advice from the specialist metabolic team throughout and it is clear who is taking forward the actions agreed) The local designated paediatrician will contact the family on the same day that the screening result is available to inform them of the positive MSUD screening result. The family must be provided with the following information:- • Outline plan for **urgent hospital** admission and assessment at local hospital (this will be to the appropriate hospital that has 24 hour paediatric cover) and subsequent inter-hospital transfer to specialist hospital or admission to specialist hospital. Ambulance transport can be arranged if required. • 'MSUD is suspected 'leaflet via email or web link • Contact numbers for the specialist metabolic team

First family face to face review with paediatrician (including confirmatory tests and clinical evaluation)	This should happen on the **same day** as the result is available. Local designated paediatrician (or on call Paediatric Consultant, or registrar or equivalent grade at admitted local hospital) to liaise with specialist metabolic team Management : A. Clinical assessment and admission to hospital **regardless of clinical status** B. Obtain blood gases, U&E, LFT,FBC, cultures, urine ketones dipstick(to go to local laboratory) C. Site IV cannula D. Obtain diagnostic samples: • Plasma (lithium heparin) for amino acid analysis

	• Urine (no preservative) for organic acid analysis Local paediatrician to liaise with the specialist laboratory (UHW) regarding sample transport. E. Local paediatrician to liaise with specialist metabolic centre regarding clinical status F. Commence clinical management: a. IV 10% dextrose/0.45% saline added potassium infusion b. Transfer to specialist centre. If GCS<8, intubate, ventilate and organise paediatric intensive care retrieval c. If transfer not possible the same day, specialist team to organise supplies of MSUD Anamix infant, Isoleucine and Valine sachets and feeding plan. Continue to liaison between specialist centre and local hospital until transferred. G. Continue to liaise with specialist hospital until baby is transferred. Local designated paediatrician must inform GP and send MSUD is suspected GP letter via email. The GP to inform health visitor. Pre-diagnosis management should include: • Explanation of the condition including introduction to inheritance. • Introduction to dietary management and use of Emergency Regimen for illness • Contact with specialist dietitian • Local paediatrician to ensure family have received – MSUD specialist team contact details, A&E letter, appropriate dietary and ER guidelines • Review available test results • (If at DGH, do not discharge until agreed by specialist team)

First Review with Specialist metabolic team	The specialist metabolic team must comprise of : • A consultant inherited metabolic disease paediatrician with relevant expertise • A paediatric dietician with metabolic expertise • A clinical nurse specialist with metabolic expertise If **well baby** specialist team to review within 5 working days of diagnostic sampling with the results. If **unwell baby** specialist team to review face to face within 2 working days with the available results. Arrange to feedback remaining results by 5 working days of diagnostic sampling. If MSUD diagnosis not confirmed on 1st visit, confirm if intermittent MSUD or further investigations required (to exclude liver disease including galactosaemia)

Upon confirmation of diagnosis	Dietary management and when to implement emergency regimen must be emphasised. Post diagnosis discussion should ensure that parents have good understanding of the condition, support information, correct contact details for specialist metabolic team, information for A&E, age appropriate dietary management/ emergency regimen information. A specialist nurse should be available to provide advice and support. The specialist dietitian should make contact with the local dietitian if appropriate. Older siblings to be tested as appropriate. Families to be made aware of opportunities for early postnatal testing of subsequent siblings. Parents should be given the opportunity to have ongoing access to a specialist dietitian and should be provided with appropriate contact details.