Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) 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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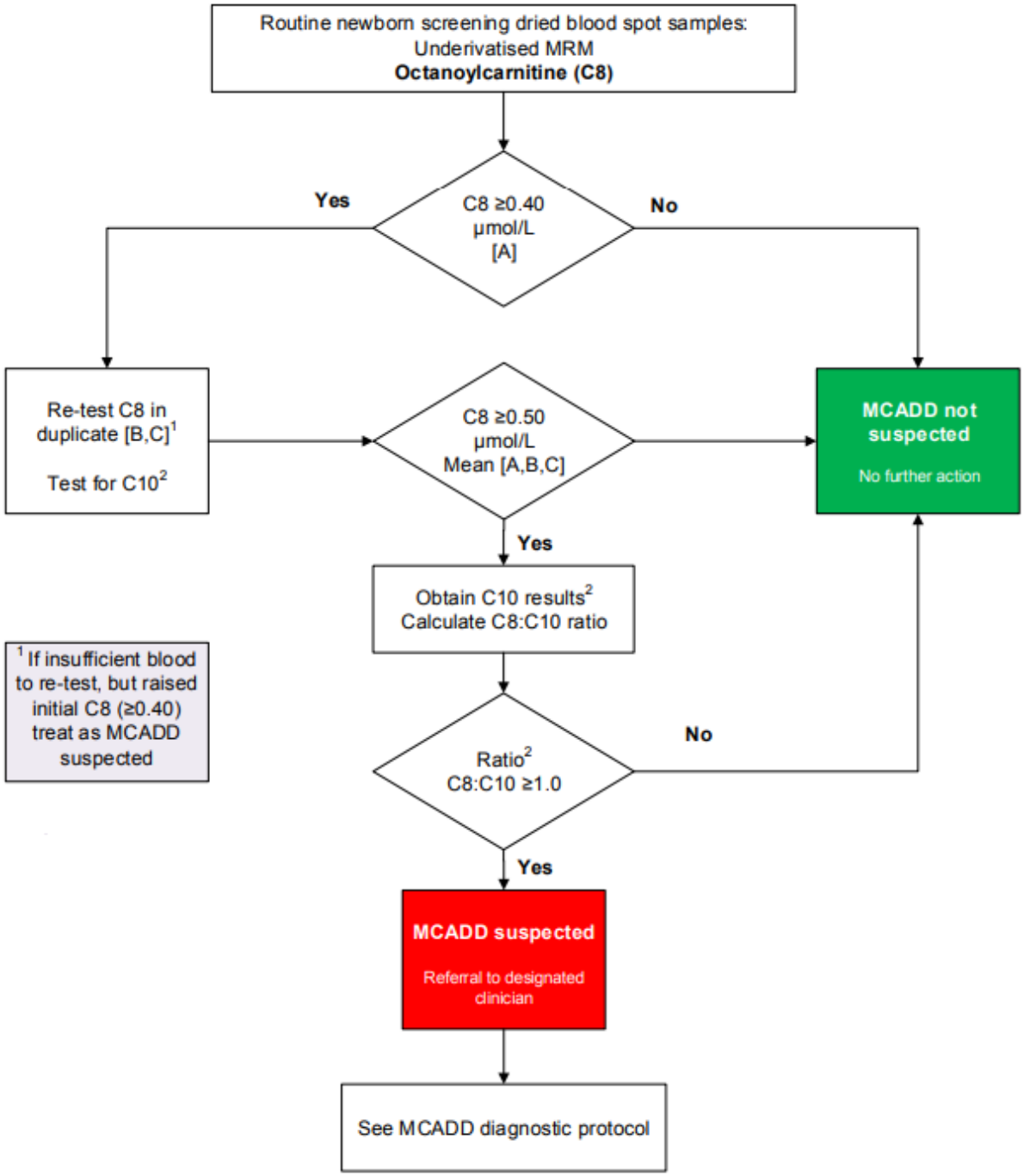
MCADD newborn screening protocol



Stage of Process	Guidelines
Defining an MCADD suspected screening result:	If a sample from a baby is found to have an octanoylcarnitine (C8) concentration equal to or greater than 0.40 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 0.5 µmol/L AND the mean C8:C10 ratio is equal to or greater than 1.0, this is a presumptive positive screening result.
Referral of babies with a positive ('MCADD 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 to Cardiff and Vale UHB 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. • 'MCADD is suspected' leaflet • MCADD GP letter • MCADD A&E letter • MCADD Dieticians letter • MCADD referral guidelines Newborn Screening Wales If a disorder other than MCADD 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 MCADD screening result. The family must be provided with the following information:- • 'MCADD is suspected' leaflet via email or web link. • Contact numbers for specialist metabolic team • Time and location of appointment

First family face to face review with paediatrician (including confirmatory tests and clinical evaluation)	This should happen on the **same** day or the **next day** 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 for assessment. Confirmatory diagnostic samples should be collected at this face to face visit (i.e. within 24 hours of receiving the screening result). Consent for DNA testing to be obtained. Specimens for the following tests should be collected: • Repeat blood spot or lithium heparin plasma for acylcarnitines: C8 duplicate and full scan • Urine (no preservative) for qualitative organic acids analysis • Two blood spot cards for DNA analysis (c.985>G and Extended Mutation Screening. Card 1 sent immediately for common mutation analysis, card 2 held by the metabolic diagnostic laboratory until the c.985>G results have been obtained). Pre-diagnosis management should include: • Explanation of the condition including introduction to inheritance • Introduction of dietary management (including maximum safe fasting times) and use of emergency regimen for illness • Contact with specialist dietician • Ensure the family have received available written information 'MCADD is suspected' leaflet and 'MCADD A&E letter'. • Contact numbers for the MCADD specialist team Local designated paediatrician to inform GP, send MCADD GP letter via email. The GP to inform health visitor.

Follow up visit with specialist metabolic team contact	An MCADD specialist 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 The follow up visit must be undertaken with the specialist MCADD team within **5 working days** of the first face to face review with the local paediatrician. To discuss results of confirmatory tests.
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 MCADD specialist 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.