Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Glutaric aciduria type 1 (GA1) 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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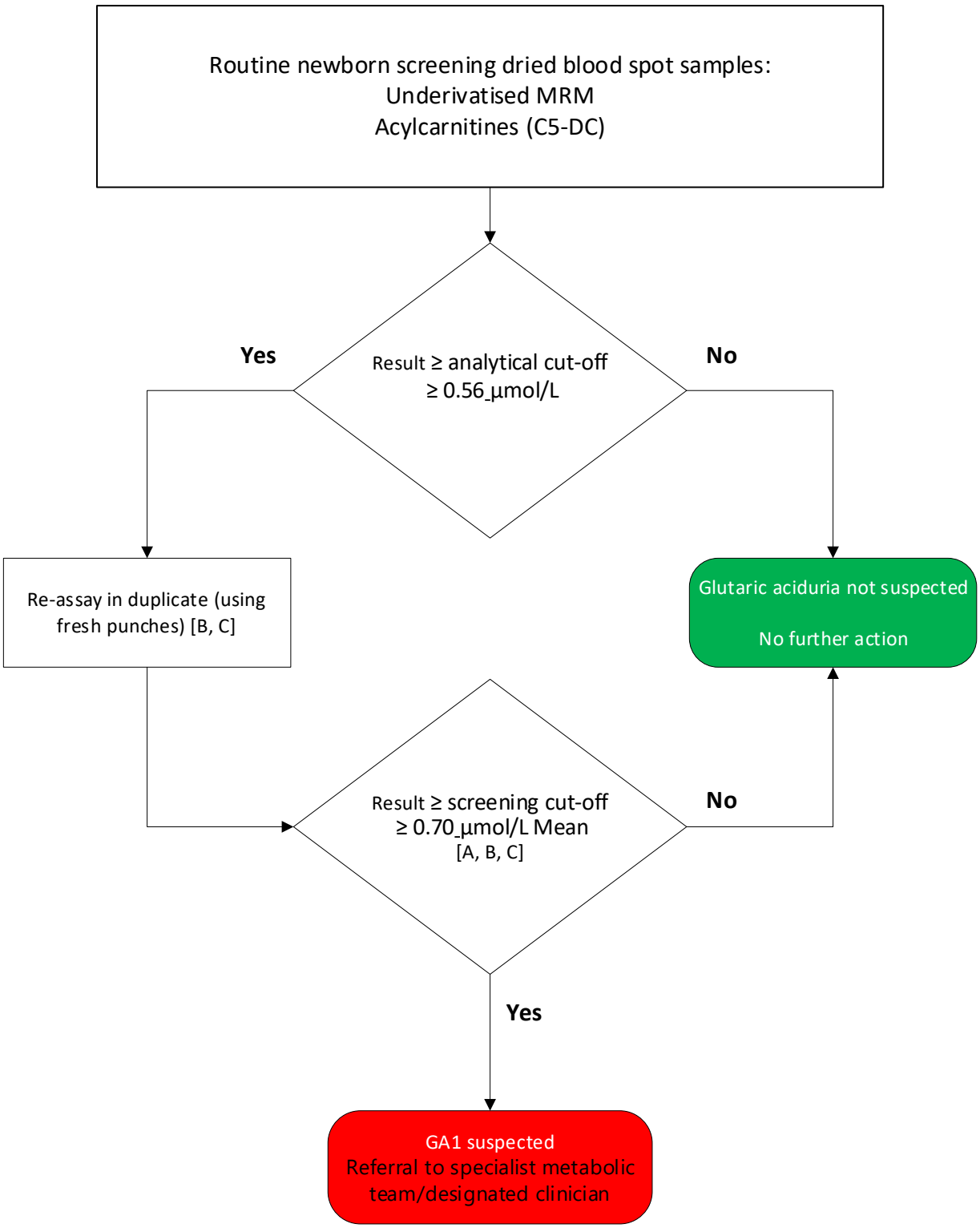
GA1 newborn screening protocol



Stage of Process	Guidelines
Defining a GA1 suspected screening result:	If a sample from a baby is found to have a C5-DC concentration equal to or greater than 0.56 µmol/L, a repeat test should be performed in duplicate on the original bloodspot card. If the mean of triplicate results is equal to or greater than 0.70 µmol/L this is a presumptive positive screening result.
Referral of babies with a positive ('GA1 suspected') screening result:	**Betsi Cadwaladr UHB referrals will be to the Manchester specialist metabolic team. For all other health boards in Wales 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. 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. • 'GA1 is suspected' leaflet • GA1 A&E letter If a disorder other than GA1 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 to inform them of the positive GA1 screening result. The family must be provided with the following information:- • Outline plan for assessment at local hospital (this will be to the appropriate hospital that has 24 hour paediatric cover). Discussion with specialist metabolic team on appropriate management of individual situation to take into account current health status of baby. • 'GA1 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 for assessment. Commence clinical management: **Well baby** A. Ensure adequate feeding B. Discharge home with BIMDG emergency guidelines and glucose polymer. Instruct to take to hospital if unwell (when baby is discharged). C. Take diagnostic samples (consent for DNA testing to be obtained): • Blood spot card for C5-DC and full acylcarnitine scan • Urine (no preservatives) for organic acid analysis • Dried blood spot card for DNA analysis Local designated paediatrician to liaise with the specialist laboratory (UHW) regarding sample transport. **Unwell Baby** A. Hospital to inform the specialist centre when baby arrives B. Obtain diagnostic samples (consent for DNA testing to be obtained) • Blood spot card for C5-DC and full acylcarnitine scan • Urine (no preservatives) for organic acid analysis • Dried blood spot card for DNA analysis Send urgently to specialist centre laboratory by courier. Inform laboratory to expect samples C. Ensure adequate feeding D. 10% Dextrose infusion +Carnitine if unwell. E. Reintroduce natural protein within 24-48 hours (refer to dietetic management pathway and advice from specialist dietitian). Local designated paediatrician to inform GP, send GA1 GP letter via email. The GP to inform the health visitor. Pre-diagnosis management should include: • Explanation of the condition

	• Ensure family have received – specialist metabolic team contact details • Ensure family have received available written information including 'GA1 is suspected' leaflet • Ensure family have received GA1 A&E letter • Introduction to dietary management and use of Emergency regimen for illness.

First Review with Specialist metabolic team	The specialist metabolic team must comprise of : • A consultant inherited metabolic disease paediatrician with relevant expertise • A paediatric dietitian with metabolic expertise • A clinical nurse specialist with metabolic expertise Within **5 working days** of diagnostic sample: Review available results and communicate result to family. If biochemistry **abnormal** : Arrange 1st follow up visit within **1 working day** of communication with family. If biochemistry **normal** : 1st follow up visit – with DNA results within **15 working days** of the diagnostic sample.

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 GA1 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 dietician 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.