Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Isovaleric Acidaemia (IVA) 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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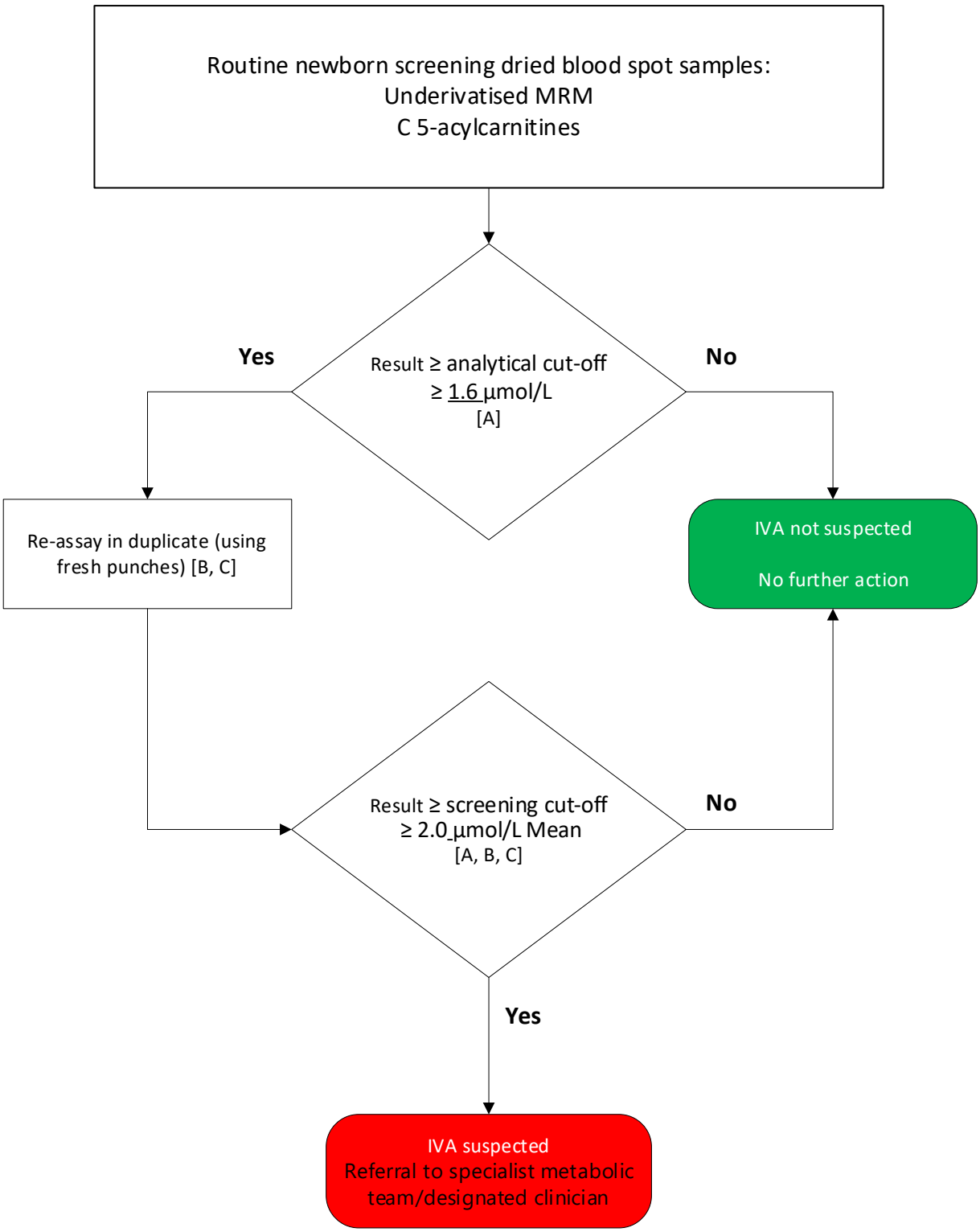
IVA newborn screening protocol



Stage of Process	Guidelines
Defining an IVA suspected screening result:	If a sample from a baby is found to have a C5 acylcarnitine equal to or greater than 1.6 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 2.0 µmol/L, this is a presumptive positive screening result.
Referral of babies with a positive ('IVA 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. • 'IVA is suspected 'leaflet • IVA A&E letter • IVA GP letter If a disorder other than IVA 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 to inform them of the positive IVA screening result. The family must be provided with the following information:- • Outline plan for urgent admission and 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, locally available healthcare provision and if child needs to be transferred to specialist centre. Ambulance transport can be arranged if required. • 'IVA 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. Obtain diagnostic samples (consent for DNA testing to be obtained): • Dried blood spot card for C5 and full acylcarnitine scan • Urine (no preservative) for organic acid analysis • Dried blood spot card for DNA analysis Send urgently to specialist centre laboratory C. Discharge home with BIMDG emergency guidelines and glucose polymer. Instruct to take to hospital if unwell (when baby is discharged). **Unwell baby** A. Clinical assessment and admission to hospital regardless of clinical status. B. Obtain blood gases, glucose, ammonia, U&E, LFT, FBC, cultures, urine ketones dipstick (samples to go to the local laboratory). Site IV cannula. C. Hospital to liaise with specialist team regarding clinical status. D. Obtain diagnostic samples (consent for DNA testing to be obtained): • Dried blood spot card for C5 and full acylcarnitine scan • Urine (no preservative) for organic acid analysis • Dried blood spot card for DNA analysis Send urgently to specialist centre laboratory (courier) E. IV 10% dextrose infusion F. Carnitine - specialist team to organise supply and send to local hospital if necessary. G. Reintroduce natural protein within 24-48 hours (refer to dietetic management pathway and advice from specialist

	dietician) H. Transfer to specialist centre as soon as appropriate Local designated paediatrician to liaise with diagnostic laboratory- inform lab to expect samples (including transport arrangement) and which hospital child has gone to in case samples need following up. Local designated paediatrician to feedback to specialist metabolic team with a review within 2 hours of admission if not already transferred to specialist centre. Local designated paediatrician to inform GP, send IVA GP letter via email. 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 'IVA is suspected' leaflet • Ensure family have received IVA 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 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.

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 dietician and should be provided with appropriate contact details.