Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Phenylketonuria (PKU) 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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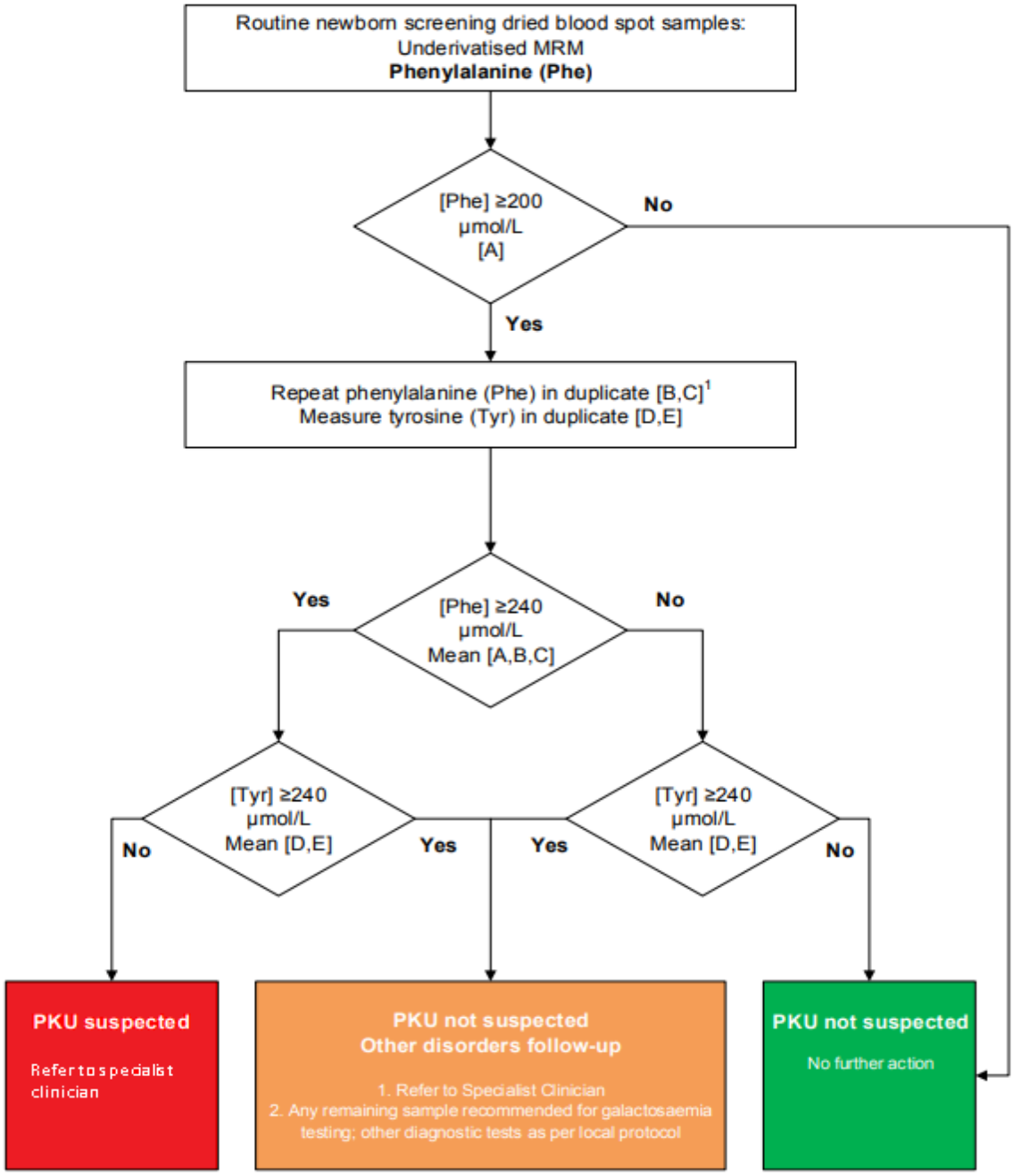
PKU newborn screening protocol



If insufficient blood to re-test and [Phe] ≥200 □mol/L arrange urgent referral to specialist clinical team

Stage of Process	Guidelines
Defining a PKU suspected screening result:	If a sample from a baby is found to have a phenylalanine concentration equal to or greater than 200 µmol/L, a repeat phenylalanine test and initial tyrosine test should be performed in duplicate on the original bloodspot card as soon as possible and within one working day. If the mean of the phenylalanine results are equal to or greater than 240 µmol/L (and mean of tyrosine <240 µmol/L) this should be reported as a 'PKU suspected' screening results. Other disorder is suspected if tyrosine is increased.
Referral of babies with a positive ('PKU suspected') screening result:	**Betsi Cadwaladr UHB referrals will be to Manchester specialist metabolic team. For all other health boards in Wales, PKU 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. Note: For those babies with a screen positive result reported on a Friday, provision must be made to see the family face to face on the same day (Friday) or the next day as necessary. If a disorder other than PKU is suspected referral must be made to the specialist metabolic team.

First family contact	The local designated paediatrician will contact the family on the day that the screening result is available to inform them of the positive PKU screening result. And arrange to see the family the **same day or next day.** The family must be provided with the following information:- • 'PKU is suspected 'leaflet via email or web link • Contact numbers for the specialist metabolic team • The time and location for the appointment

First family face to face review with paediatrician (including confirmatory tests)	Pre-diagnosis management should include: • Explanation of the condition • Ensure family have received the specialist metabolic team contact details • Ensure family have received available written information including 'PKU is suspected' leaflet • Explanation of dietary management and monitoring (if clinically required). When a baby is seen with a positive screening result, specimens for the following tests should be collected that day to confirm diagnosis and exclude other possible metabolic disorders: • Venous blood (lithium heparin) for quantitative plasma amino acids • Two blood spot cards with non-anticoagulated blood for biopterin analysis. • These samples can also be used to exclude/confirm other disorders that may be associated with raised phenylalanine, including galactosaemia, tyrosinaemia and biopterin defects where appropriate. **Detection of these disorders is not the objective of the PKU screening programme and all such cases will not be reliably detected.** Results of confirmatory phenylalanine test to be available within one working day of specimen collection. Diet should be commenced on this initial visit if clinically appropriate. Arrangements should be made for follow-up blood phenylalanine monitoring samples to be collected. The local paediatrician to inform GP and send PKU is suspected GP letter via email. GP to inform health visitor.

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 Follow up management should include: • A review of the condition and inheritance, supported with written information • Review of dietary management, growth and development and general health • Support information – including contact details of parent support organisations • Parents should be taught how to perform the heel-prick on their baby • Support from a specialist nurse should be available to provide advice and support if needed • Specialist dietician to make contact with local dietitian as appropriate • Older siblings to be tested as appropriate. Families to be made aware of opportunities for early postnatal testing of subsequent siblings.

Upon confirmation of diagnosis	Post diagnosis discussion should ensure that parents have good understanding of the condition and inheritance, supported with written information. Discussion on dietary management, blood monitoring requirements, growth and development and general health. Families should be taught how to perform the heel prick on their baby. A specialist nurse should be available to provide advice and support. The specialist dietitian should make contact with the local dietitian if appropriate. Families to be made aware of opportunities for early postnatal testing of subsequent siblings. Contact details of parent support organisations