Regional Policy template for newborn screening for Inherited Metabolic Disease (IMD) in the South West (Bristol Newborn Screening Laboratory)

Produced by the SW Regional IMD Implementation Group
August 2015

Participating Trusts:

North Bristol NHS Trust
North Devon NHS Trust
Gloucestershire Care Services NHS Trust
Gloucestershire Hospitals NHS Foundation Trust
Plymouth Hospitals NHS Trust
Royal Cornwall Hospitals NHS Trust
Royal Devon and Exeter NHS Foundation Trust
Taunton and Somerset NHS Foundation Trust
Torbay and South Devon NHS Foundation Trust
University Hospitals Bristol NHS Foundation Trust
Weston Area Health NHS Trust
Yeovil District Hospital NHS Foundation Trust
## Policy history

<table>
<thead>
<tr>
<th>Version No.</th>
<th>Date</th>
<th>Author</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>20/11/2014</td>
<td>Helena Kemp</td>
<td>Distributed Dec 14</td>
</tr>
<tr>
<td>2</td>
<td>29/7/2015</td>
<td>Helena Kemp</td>
<td>Updated to reflect national guidelines and pathways ratified by IMD</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Screening Advisory Board April 28 2015</td>
</tr>
<tr>
<td>3</td>
<td>21/8/15</td>
<td>Helena Kemp</td>
<td>Links updated due to changes in screening websites and transfer of</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>all screening information to Gov.UK website</td>
</tr>
<tr>
<td>4</td>
<td>21/12/15</td>
<td>Helena Kemp</td>
<td>Added names of participating Trusts</td>
</tr>
</tbody>
</table>
Contents

Introduction ................................................................................................................................. 3
1 The conditions .......................................................................................................................... 4
2 Aims ......................................................................................................................................... 8
3 Objectives ............................................................................................................................... 9
4 The screening protocols ......................................................................................................... 10
  4a The screening process ........................................................................................................ 11
  4b Process for offer and taking of test ...................................................................................... 15
  4c Late testing .......................................................................................................................... 15
  4d Sibling testing ...................................................................................................................... 15
5 Newborn screening results ..................................................................................................... 16
  5a Condition ‘not suspected’ .................................................................................................. 16
  5b Condition ‘suspected’ .......................................................................................................... 17
  5c Clinical referral pathways

6 Training and education ........................................................................................................... 26
7 Clinical Governance and Quality Management ..................................................................... 27
8 Regional contacts .................................................................................................................... 1
9 Local contacts ........................................................................................................................ 2
10 References
Introduction

Screening for inherited metabolic disease has been in place in the UK since the 1960’s when screening for Phenylketonuria (PKU) first began. More recently screening for Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD) was introduced and in 2012/13 a study was undertaken to evaluate the utility for screening for other additional IMDs. In May 2014 the UK National Screening Committee announced its recommendation to include four additional inherited metabolic diseases as part of the national newborn screening blood spot programme, maple syrup urine disease (MSUD), isovaleric aciduria (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU),

The South West Regional Policy (Bristol Newborn Screening Laboratory) has been produced by the SW Regional IMD Implementation Group as a guide for local implementation teams to inform the introduction of newborn screening for the four additional conditions throughout the region. At the same time the regional policies for PKU and MCADD have been reviewed and, where possible, aligned to enable the production of a single unified policy for screening for IMD across the SW Region.

The early detection offered by newborn screening is a significant benefit for patients with all of the conditions included within the programme and in some cases can be lifesaving. The successful introduction of screening depends upon the cohesive management of testing as a programme of care from pre-screening information to screening results and in the case of positive cases enrolment into appropriate treatment. It is important that mothers are fully informed at each stage and that only clinically significant disease is detected with a minimum number of false positive cases. Once a screen positive patient is identified, the period of uncertainty required for any associated confirmatory testing must be kept to a minimum and parents supported during this trying time.

The policy has been developed in line with the policies and standards of the National Blood Spot Screening Programme and UK Newborn Screening Programme Centre. Further information, along with the National Programme Policies, Standards and resources can be found on the website https://www.gov.uk/topic/population-screening-programmes
1 The conditions

Phenylketonuria

Phenylketonuria is one of the most common inherited metabolic disorders with an incidence across the UK of approximately 1 in 10,000 births. PKU is an autosomal recessively inherited disorder of amino acid metabolism caused by a deficiency of the enzyme phenylalanine hydroxylase (PAH). The enzyme is required to metabolise phenylalanine to tyrosine. A deficiency of the enzyme results in an accumulation of phenylalanine and associated metabolites in blood and tissues. The infant brain is sensitive to high phenylalanine levels and if left untreated, patients with PKU develop severe mental retardation, microcephaly and a proportion of patients develop epilepsy. Older patients have behavioural problems and some suffer from psychiatric illness.

Children with PKU requiring treatment are treated with a phenylalanine restricted diet to reduce the flux through the affected metabolic pathway thereby preventing the accumulation of toxic metabolites. Dietary therapy utilises a phenylalanine-free synthetic amino acid mixture as a substitute for natural protein and requires careful management to ensure appropriate vitamins and trace elements together with small amounts of natural protein are provided. Treatment should begin with minimum delay. Recommendations in the past have indicated that diet should be initiated by 20 days of age and continued throughout childhood.

An increased blood phenylalanine is not specific for PKU. In addition to disorders of phenylalanine hydroxylase deficiency, increased phenylalanine on newborn screening at 5-8 days may occur in several other situations. There are several rare disorders of pterin synthesis/recycling which may be associated with an isolated increase in phenylalanine at screening and therefore detected by the PKU programme. All babies with a PKU suspected screening result are tested for pterin disorders at follow up.

In some cases the phenylalanine increase is associated with an increase in tyrosine which is measured as part of the screening protocol. This situation can arise in conditions which cause liver disease in the neonate e.g. hepatitis, biliary atresia, CMV and some inherited disorders, in particular Galactosaemia and Tyrosinaemia type 1. There are also a number of other non-specific causes including prematurity, parenteral nutrition and transient illness. Babies with an associated increase in tyrosine require different investigation and management and urgent referral to an appropriate specialist clinician is essential.

Medium Chain Acyl CoA Dehydrogenase Deficiency

MCADD is an autosomal recessive condition that affects approximately 1 in 10,000 born in the United Kingdom. It is caused by a deficiency of an enzyme medium chain acyl CoA dehydrogenase. This enzyme is required to breakdown certain stored fats, medium chain length fatty acids, and is necessary to enable the body to use its own fat reserves to produce energy in periods of prolonged fasting or stress.

Complications of MCADD can arise during prolonged fasting but particularly occur during intercurrent illness when the body's demand for energy is high and Calorie
intake is often reduced. As fat fuels cannot be used these patients quickly use up glucose and can develop hypoglycaemia. Partially broken down fatty acids accumulate and this, along with low blood glucose concentrations, cause these patients to become drowsy, comatose and eventually to stop breathing. Without treatment this condition is associated with significant morbidity and mortality. Early identification by screening however results in early pre-symptomatic treatment reducing the risk of acute life-threatening episodes.

MCADD requires no special treatment when the child is well. Fasting should however be avoided. When illness occurs MCADD is easily treatable with an emergency feeding regime of high calorie drinks or, if unable to tolerate e.g. due to vomiting, urgent hospital admission for an intravenous 10% dextrose infusion. With early detection, monitoring and avoidance of fasts, children diagnosed with MCADD can lead normal lives particularly as ‘safe’ times between meals increase as they grow older.

**Maple Syrup Urine disease**

MSUD is an autosomal recessive caused by a deficiency of the branched chain alpha keto acid dehydrogenase complex. The condition occurs in approximately 1:200,000 live births. The resulting metabolic block leads to an increased concentration of the branched chain amino acids leucine, valine and isoleucine and their corresponding keto acids. These compounds accumulate in tissues resulting in a life threatening metabolic decompensation in some affected individuals and are elevated in the blood and urine. The name of the condition derives from the sweet smelling urine sometimes produced by affected individuals which some have likened to the smell of maple syrup.

The classic form of the disorder presents shortly after birth often in the first two weeks of life. Vomiting or difficulty feeding are often early symptoms accompanied by lethargy and progressive neurological deterioration. Intermediate and intermittent forms of the condition are also described. Patients with the intermediate form may present with developmental delay although the characteristic elevation of branched chain amino acids is still present. The intermittent form of the disease may only manifest at times of stress or infection and branched chain amino acids may not be continuously elevated. Rarer thiamine responsive disease has been described together with another variant which also affects the pyruvate dehydrogenase complex resulting in marked lactic acidosis.

It is likely that newborn screening will detect patients with the classic condition but may not detect individuals with intermediate or intermittent forms which have a spectrum of clinical and biochemical severity.

**Isovaleric aciduria**

Isovaleric acidaemia (IVA) is caused by a deficiency of isovaleryl-CoA dehydrogenase (IVD), an enzyme involved in the catabolism of the amino acid leucine. It is an autosomal recessive disease, with an estimated incidence of around 1 in 100,000 with a higher incidence in some locations and ethnic groups.

Loss of function of the enzyme leads to the toxic build-up of metabolites including isovaleric acid and its glycine and carnitine derivatives. The disease has a spectrum of clinical phenotypes which might include acute neonatal presentations, acute presentations at a later age and chronic intermittent presentations. The acute neonatal presentation is characteristically in the first two weeks after birth. Infants
are initially well, then develop vomiting and lethargy, progressing to coma. Patients may also present with similar symptoms at a later age, usually precipitated by an infection. Other patients present with chronic symptoms - failure to thrive and/or developmental delay, usually within the first year.

Over 25 mutations in the IVD gene have been associated with disease, a number of which lead to complete lack of the enzyme. Although a firm phenotype/genotype correlation has not been identified, recent research suggests that the 932C>T mutation in the IVD gene may be associated with a milder phenotype. Newborn screening has identified individuals with partial as well as complete IVD deficiency.

**Glutaric aciduria type 1**

Glutaric aciduria type 1 (GA1) is an autosomal recessive condition caused by a deficiency of the enzyme glutaryl-CoA dehydrogenase (GCDH). The estimated incidence in the UK is around 1 in 100,000 live births.

GCDH is involved in the metabolism of the amino acids lysine, hydroxylysine, and tryptophan. Defective catabolism causes the toxic accumulation of glutaric acid, 3-hydroxyglutaric acid, glutaconic acid, and glutaryl carnitine.

GA1 frequently presents with an encephalopathic crisis, which is most commonly between 9 months to 2 years. These are usually precipitated about 1-3 days after onset of a non-specific intercurrent illness, gastrointestinal infection or pneumonia and lead to dystonia and dyskinesia as permanent sequelae but with relative preservation of the intellect. There are also two other clinically recognised subgroups of patients with GA1 who have an insidious or later onset form with non-specific symptoms including irritability and transient lactic acidosis. These infants go on to show delayed motor development. Very occasionally an individual might be asymptomatic.

Over 150 disease causing mutations have been identified; of these the R402W mutation is the most prevalent among Caucasians. Most mutations, including the R402W mutation, are associated with undetectable GCDH activity and excretion of high amounts of glutaric acid. However, mutations that lead to varying levels of residual GCDH activity and low excretion of glutaric acid have also been reported. Consequently, patients with GA1 can be divided into two biochemically defined subgroups based on the levels of glutaric acid present in the urine: low excretors and high excretors. Although these subgroups are clinically similar it is important to be aware as confirmatory testing in low excretors requires more complex follow-up, with either determination of GCDH enzyme activity or by mutation analysis of the GCDH gene.

**Homocystinuria**

The most common cause of homocystinuria is a defect in the enzyme cystathionine \( \beta \)-synthase (CBS); this is referred to as “classical” homocystinuria. The overall incidence in the UK is reported to be around 1 in 100,000 live births. The mode of inheritance of classical homocystinuria is autosomal recessive.

Classical homocystinuria is associated with a number of clinical and pathological abnormalities. Infants are usually normal at birth and without screening the diagnosis is not usually made until the first 2-3 years of life. Myopia followed by dislocation of the lens, osteoporosis, thinning and lengthening of the long bones,
mental retardation and thromboembolism affecting larger and small arteries and veins are the commonest clinical features. Without treatment, 25% of patients will die before the age of 30, usually as a result of arterial thromboembolism. There is a great deal of clinical heterogeneity, with some patients displaying all clinical symptoms whilst others display very few or none. The concentration of plasma total homocysteine can be measured to assess the clinical severity of disease and can be monitored to determine the response to treatment.

Homocystinuric patients can be sub-divided into two important biochemical phenotypes:

- Pyridoxine responsive (screen undetectable)
- Pyridoxine non-responsive (screen detectable)

In the UK approximately 50% of patients with classical homocystinuria are classified as pyridoxine responsive, these patients usually have milder symptoms and disease progression is slower and slowed further by oral pyridoxine (Vitamin B6) supplementation, they are very unlikely to be detected by newborn screening
2  Aims

1. To offer screening to all babies resident within the area covered by the Bristol Newborn Screening Laboratory

2. To inform parents of the IMD status of their newborn children.

3. To minimise morbidity and mortality from PKU, MCADD, MSUD, IVA, GA1 and HCU in infants born within the South West region.
3 Objectives

1. To identify and offer screening to all eligible infants.
2. To process tests for all those screened in a timely manner.
3. To ensure an appropriate level of understanding amongst professionals involved with the programme.
4. To appropriately follow up all infants identified as needing further investigative testing.
5. To accurately diagnose all infants born with PKU, MCADD, MSUD, IVA, GA1 and HCU.
6. To ensure that information is provided to affected families in an acceptable and accessible manner and is appropriate to the needs of the relevant communities.
7. To ensure effective and acceptable follow up, care and support for infants with PKU, MCADD, MSUD, IVA, GA1 and HCU and their carers.
8. To audit the service on an ongoing basis and provide information on acceptability, effectiveness and resource implications.
9. To minimise the adverse effects of screening (anxiety, misunderstanding, inaccurate information, unnecessary investigation and follow-up, and inappropriate disclosure of patient-specific information).
The newborn screening protocols have been agreed and endorsed by the IMD Scientific Advisory Board. The screening protocols are intended to:

**Maximise** early detection of PKU, MCADD, MSUD, IVA, GA1 and HCU so that appropriate early pre-symptomatic treatment can be initiated and the risk of acute, life threatening episodes and/or the development of disease associated complications can be reduced.

**Minimise** the number of false positive results and the detection of unaffected carriers.

Samples will be processed at a laboratory recognised as a newborn blood spot screening laboratory, adhering to the programme laboratory standards and using the recommended standard operating procedures: [https://www.gov.uk/topic/population-screening-programmes](https://www.gov.uk/topic/population-screening-programmes)

Note screening for IMD will include all 6 conditions. Patients can only consent or decline all 6 conditions.
## 4a The screening process

<table>
<thead>
<tr>
<th>Policy</th>
<th>Local arrangements</th>
<th>Professional responsible</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Local policy and practice for service delivery for all aspects of the programme (including laboratory and diagnostic services) must adhere to national newborn blood spot screening standards and guidance, August 2013 <a href="https://www.gov.uk/topic/population-screening-programmes">https://www.gov.uk/topic/population-screening-programmes</a></td>
<td>[insert local arrangements]</td>
<td></td>
</tr>
<tr>
<td>2. All women will be given a copy of the pre-screening information leaflet during the third trimester to enable parents to make an informed choice based on their individual values</td>
<td>[insert local arrangements]</td>
<td><em>[e.g. community midwife]</em>.</td>
</tr>
<tr>
<td>3. Screening should be preceded by the provision of written information, where possible in the parent’s first language. There should be access to additional information and support if requested</td>
<td>At least 24 hours before taking the heel prick, the midwife should ensure parents have a copy of the pre-screening information leaflet. This is available electronically via <a href="https://www.gov.uk/topic/population-screening-programmes">www.gov.uk/topic/population-screening-programmes</a> and is included within ‘Newborn Blood Spot Screening – Information and Support for Parents’.</td>
<td>* [e.g. community midwife].</td>
</tr>
</tbody>
</table>
| 4. Screening for IMD (includes PKU, MCADD, MSUD, IVA, GA1 & HCU) will be offered on a resident basis, at ideally 5 days of age, as part of the Newborn Bloodspot Screening Programme. **Standard 4**  
**Acceptable Standard**  
95% of first samples taken 5-8 days after birth (ideally day 5).  
**Achievable Standard**  
99% of first samples taken 5-8 days after birth (ideally day 5). | [insert local arrangements] | * [e.g. community midwife]. |
5. The offer of screening and the decision to accept or decline should be recorded. The blood spot card should be completed and clearly marked ‘DECLINE’ and sent to the screening laboratory.

**Note a decline for IMD screening includes all 6 conditions. It will not be possible to decline IMD screening for one or more of the 6 conditions.**

<table>
<thead>
<tr>
<th>Policy</th>
<th>Local arrangements</th>
<th>Professional responsible</th>
</tr>
</thead>
<tbody>
<tr>
<td>5.</td>
<td>[Insert detail of local arrangements and method of documentation e.g. recording in hand-held maternity notes and in the red book and blood spot card to be sent to screening laboratory for declines].</td>
<td>* [e.g. community midwife].</td>
</tr>
</tbody>
</table>

6. Ensure the good quality blood spot is collected i.e. sample is taken at the right time, all data fields completed, contains sufficient blood for all tests, is not contaminated and reaches the lab in a timely manner.

**Standard 6**
**Acceptable standard**
Avoidable repeat rate ≤ 2%

**Achievable standard**
Avoidable repeat rate < 0.5%

<table>
<thead>
<tr>
<th>6.</th>
<th>Insert local mechanisms to optimise and monitor avoidable repeat rate including KPI reporting and monitoring</th>
</tr>
</thead>
</table>

7. The NHS number must be used on the blood spot card to identify the baby preferable provided on a bar-coded label.

**Standard 3**
**Acceptable Standard**
100% of blood spot cards received by a laboratory include the babies’ NHS Number.

**Achievable Standard**
95% of blood spot cards received by laboratory should have a bar-coded label including the babies’ NHS number.

<table>
<thead>
<tr>
<th>7.</th>
<th>[insert local arrangements]</th>
</tr>
</thead>
<tbody>
<tr>
<td>8.</td>
<td>[insert local arrangements]</td>
</tr>
</tbody>
</table>

**NB Dispatch of blood spot screening cards should not be delayed in order to batch cards**

<table>
<thead>
<tr>
<th>8.</th>
<th>[insert local arrangements]</th>
</tr>
</thead>
</table>

8. Blood spot screening cards should be sent to the screening laboratory as soon as possible.

<table>
<thead>
<tr>
<th>8.</th>
<th>[insert local arrangements]</th>
</tr>
</thead>
</table>

**NB Dispatch of blood spot screening cards should not be delayed in order to batch cards**

<p>| 8. | [insert local arrangements] | * [e.g. community midwife]. |</p>
<table>
<thead>
<tr>
<th>Policy</th>
<th>Local arrangements</th>
<th>Professional responsible</th>
</tr>
</thead>
<tbody>
<tr>
<td>possible after the sample has been taken</td>
<td>together in one envelope..</td>
<td></td>
</tr>
<tr>
<td><strong>Standard 5</strong>&lt;br&gt;Acceptable Standard&lt;br&gt;&gt;99% of samples received by laboratory within 4 working days of blood sample being taken.&lt;br&gt;<strong>Achievable Standard</strong>&lt;br&gt;&gt;99% of samples received by laboratory within 3 working days of blood sample being taken.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>9. Professional responsibilities to support women and their partners throughout the screening process and manage adverse incidents to be identified in the local care pathway.</td>
<td>See care pathway.</td>
<td>Professional responsibility is identified in the care pathway.</td>
</tr>
</tbody>
</table>
| 10. There should be explicit governance arrangements to ensure all babies, including movers in, are offered screening including a documented failsafe policy<br>**Standard 1a & 1b**<br>Acceptable &gt; 95% of eligible babies are tested for all conditions<br>Achievable &gt; 99% of eligible babies are tested for all conditions | [insert local arrangements/local failsafe policy ref e.g.  
- arrangements for offering screening to babies in neonatal units or children’ ward  
- Screening laboratory inform CHRD of status 01 codes  
- CHRD have mechanisms to notify midwifery service of babies who have not been screened by Day 17 (ideally Day 14) or within 21 days of moving into area  
- Use of the Northgate Failsafe system | Professional responsibility is identified in the care pathway. e.g.  
- Mother’s named midwife is responsible for checking that appropriate arrangements have been made  
[lab to identify person with this responsibility]  
[CHRD to identify person with this responsibility]  
CHRD to send written confirmation to GP/HV  
HCP performing 6–8 week infant physical examination to ensure result available and recorded |
Regional Policy for IMD screening.
4 b  Process for offer and taking of test

4c  Late testing

Babies who have not been screened during the newborn period should be screened up to 12 months of age in line with NHS Newborn Blood Spot Screening Programme guidance. After 12 months of age, if the family or GP have any clinical concerns a referral for paediatric assessment would be appropriate.

4d  Sibling testing

A new sibling born to the same parents when an index case with either PKU, MCADD, MSUD, IVA, GA1 or HCU has already been identified has a 1 in 4 risk of having the same disorder. In these circumstances it is recommended practice to test earlier than the 5-8 day time frame to avoid delays in diagnosis and parental anxiety. This does not remove the need to test at 5-8 days to screen for all other disorders tested as part of the blood spot screening programme.

The decision about when and how to test depends upon the disorder suspected. Note it is most important for disorders such as MSUD, MCADD and IVA which can potentially have an early neonatal presentation.

Older siblings of newly diagnosed screen detected cases, when born to the same parents, have a 1 in 4 chance of having the same disorder. Careful consideration should be given by the clinician investigating the baby referred via screening to the risks to other, older siblings in the family.

For guidance on testing of siblings contact the Regional Specialist Paediatric Metabolic Team and the Newborn Screening Laboratory.
5 Newborn screening results

All screening results will be reported to Child Health Records departments in line with present policies for the Newborn Blood Spot Screening Programme with use of the appropriate status codes.

Written policies and mechanisms for relaying of results to parents must be developed locally to include professional responsibility for informing parents of the result, and clear process detail for follow-up of affected babies. Policies should be developed in line with national and regional guidance as detailed below.

5a. Condition ‘not suspected’

Newborn screening results which are negative for all of the conditions are to be reported in a timely manner to parents.

Standard 12
Acceptable standard
100% of screen negative results letters to be dispatched direct to parents from the CHRD by 6 weeks of age
5b  Condition ‘suspected’

Babies identified as screen positive should have timely referral into clinical care in line with defined clinical referral pathway see below. Any repeat screening tests should be offered & collected in a timely manner in order that treatment and clinical referral targets are met.

### Standard 7

**Timely taking of a repeat blood spot sample**

A repeat sample from the avoidable repeat category must be taken within 3 calendar days of the receipt of the request.

### Standard 11

**Timely receipt into clinical care**

A baby in whom an IMD (on first sample) is suspected should attend their first appointment by:

- **Acceptable**: 100% by 17 days of age
- **Achievable**: 100% by 14 days of age

5c  Clinical referral and management guidelines

Guidelines for Clinical Referral for each of the 6 IMD conditions are outlined below and are in line with the guidance endorsed by the IMD Screening Advisory Board.

**Notes**

1. The Newborn Screening Laboratory (NSL) will fulfill the role of the IMD screening clinical liaison service to co-ordinate the referral of all IMD positive screens.

2. A Specialist team should comprise:
   - A consultant Inherited Metabolic Disease (IMD) paediatrician with relevant expertise.
   - A paediatric dietitian with metabolic expertise
   - A clinical nurse specialist

   A designated team must include clinicians trained to receive IMD referrals and have a paediatric dietitian.

3. Protocols, guidelines and template letters for each IMD condition (PKU, MCADD, MSUD, IVA, GA1 & HCU) are available on the newborn bloodspot screening and BIMDG websites (links included within the following clinical referral pathways):

   a. Information leaflets for parents
   b. Technical resources
      i. Screening & diagnostic & sibling testing protocols
      ii. Clinical management guidelines
      iii. Dietetic management guidelines
      iv. Emergency regimens
      v. Template letters (GP letters, A&E letter)
PKU Clinical Referral Pathway

* PKU Suspected

ON THE SAME DAY**
1. NSL informs the Specialist team who will inform the Designated team depending on babies location. First review appointment arranged (to take place within 24hrs of informing the parents). Note parents should NOT be informed of a positive result if an appointment cannot be given for the same or next day e.g. parents should not be informed on a Friday unless an appointment is for that day or the Saturday. In this case contact should be deferred until after the weekend to the next working day.
2. Specialist/Designated team CONTACT FAMILY
   - Give the family an appointment time to attend the specialist (designated) centre the same or next day (including directions as required)
   - Arrange for the family to receive the 'PKU is suspected' leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group) (this can be via email, website address or face-to-face contact as per local protocol)
   - Give the family contact numbers for the PKU specialist team
   - Give the family advice regarding feeding
3. Specialist or Designated team to:
   - Take diagnostic samples*** & send urgently to screening lab (courier). Inform lab
   - Dietetic review, commence dietary restriction if appropriate****
   - Clinician to ensure:
     a. Family have received the 'PKU is suspected' leaflet, PKU specialist (designated) team contact details and appropriate dietary guidelines
     b. Letters have been sent to the GP, local dietitian as necessary
4. NSL informs GP, send PKU GP letter via fax / email copied to HV
5. NSL informs Local Screening Co-ordinator by phone

FIRST REVIEW: FACE-TO-FACE - within 24 hours of informing family of screening result
1. Take diagnostic samples*** & send urgently to screening lab (courier). Inform lab
2. Dietetic review, commence dietary restriction if appropriate****
3. Clinician to ensure:
   a. Family have received the 'PKU is suspected' leaflet, PKU specialist (designated) team contact details and appropriate dietary guidelines
   b. Letters have been sent to the GP, local dietitian as necessary

FIRST REVIEW & COMMUNICATION OF DIAGNOSTIC RESULTS - within 15 working days of the 1st face-to-face review
1. Clinical review and results: phenylalanine, tyrosine, pterins and dihydropteridine reductase (DHPR).
2. Arrange sibling screening if diagnosis confirmed*****
3. If diagnosis not confirmed see diagnostic protocol***

PKU CONFIRMED

Follow up visits for on-going management
Dietetic and clinical review

Follow diagnostic protocol***

* PKU screening protocol see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

** PKU initial clinical management guidelines www.bimdg.org.uk

*** PKU diagnostic protocol for confirmatory tests see Laboratory Guide for IMDs https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

**** PKU Dietetic Management Pathway www.bimdg.org.uk/site/guidelines-enbs.asp

***** PKU sibling testing see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

18
ii. PKU – ‘Other Disorder Suspected’

1. For all ‘Other disorder suspected’ results the NSL will inform and discuss the result with the Specialist IMD team. The Specialist team will liaise with the relevant local paediatric team as required.

2. Same day review is recommended for this category of screening result as the findings may be indicative of a condition requiring prompt investigation and management. Review of these cases as part of the national review of PKU Screening Programme indicated that the majority of these cases were inpatients at the time of the screening result (PKU Expert Group Report http://www.newbornscreening-bloodspot.org.uk).

3. Further testing for disorders that may be associated with elevated phenylalanine and tyrosine levels should be undertaken. This will include at least full blood count, clotting, urea and electrolytes, bicarbonate, calcium, phosphate, split bilirubin, AST/ALT, alkaline phosphatase, gamma GT, albumin, cholesterol, triglycerides, urine organic acids (in particular succinyl acetone), galactose-1-phosphate uridyl transferase and plasma amino acids. If there is significant liver dysfunction advice should be sought from a liver unit.
**MCADD Clinical Referral Pathway**

*MCADD Suspected*

**ON THE SAME DAY**
1. NSL informs the Specialist team who will inform the Designated team depending on babies location. First review appointment arranged (to take place within 24hrs)
2. Specialist or Designated team to CONTACT FAMILY
3. Specialist or Designated team to**:
   - Give the family an appointment time to attend the specialist/designated) centre the same or next day (including directions as required)
   - Arrange for the family to receive the 'MCADD is suspected' leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group) and A&E letter (this can be via email, website address or face-to-face contact as per local protocol)
   - Give the family contact numbers for the MCADD specialist team
   - Give the family advice regarding feeding
4. NSL informs GP, send MCADD GP letter via fax/email & copied to HV
5. NSL informs Local Screening Co-ordinator

**FIRST REVIEW: FACE-TO-FACE - within 24 hours of screening result**
1. Consent for DNA testing to be obtained
2. Take diagnostic samples*** & send urgently to screening lab (courier). Inform lab
3. If baby well ensure adequate feeding & dietetic review & emergency regime teaching.
4. If baby unwell follow MCADD dietetic & emergency management guidelines****. Before discharge ensure adequate feeding & dietetic review/emergency regime teaching.
5. At discharge ensure family have the A&E letter, appropriate leaflet ('MCADD is suspected/confirmed), emergency regimen BIMDG Emergency guidelines glucose polymer. Instruct to take to hospital if unwell.
6. Ensure letters have been sent to the GP, local paediatrician and local dietitian as necessary

**1st FOLLOW-UP VISIT - within 5 working days of the 1st face-to-face review**
Clinical review and results: octanoylcarnitine (C8); qualitative urine organic acid (UOA); c985A>G mutation analysis
If diagnosis not confirmed – see diagnostic protocol***

**MCADD CONFIRMED**

- **No further action**
- **Yes**
  - **No**
  - **Yes**

**2nd FOLLOW-UP VISIT - within 15 working days of first follow-up visit**
Clinical review and results: extended mutation screening, quantitative UOA

**Treat as MCADD (refer to dietetic and clinical management guidelines)**
Arrange sibling screening ****

---


**MCADD initial clinical management guidelines** [www.bimdq.org.uk](http://www.bimdq.org.uk)

**MCADD diagnostic protocol** for confirmatory tests see Laboratory Guide for IMDs [https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot](https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot)


**MCADD sibling testing** see Laboratory Guide for IMDs for details [www.gov.uk/topic/population-screening-programmes/newborn-blood-spot](http://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot)
MSUD Clinical Referral Pathway

* MSUD ‘Suspected’

ON THE SAME DAY
1. NSL inform Specialist team who will inform Designated team if required (depends on babies address) to arrange urgent hospital admission
2. Specialist/Designated team to CONTACT FAMILY
3. Specialist /Designated team to instruct family to go to appropriate hospital with 24 hr paediatric cover. Offer to arrange an ambulance.
4. If initial review not at Specialist Centre, Specialist team to liaise with the hospital (Designated or on call Paediatric Consultant)
   a. Fax/email information to the hospital for clinicians and parents, MSUD A&E letter, ‘MSUD is suspected’ leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the MSUD specialist team.
   b. Clinical assessment & admission to hospital regardless of clinical status
      i. Hospital to liaise with clinical centre regarding clinical state
5. Comence clinical management** (Specialist or Designated team)
   a. Take diagnostic samples*** and send urgently to screening laboratory (courier) & inform lab
   b. IV 10% dextrose/0.45% saline + added potassium infusion
   c. Transfer to specialist centre. If GCS <8 intubate, ventilate & organise PICU retrieval
   d. If transfer not possible same day, specialist team to organise supplies of MSUD Anamix Infant, isoleucine & valine sachets and feeding plan****
      e. Continue liaison between specialist & local hospital until transferred
6. NSL inform GP, send MSUD GP letter via fax / email & copy to HV
7. NSL Informs Local Screening Midwife by phone

FIRST REVIEW within 24 hours of screening result
Ideally face to face, in exceptional circumstances via telephone/video conference/skype
Review diagnostic test results (If at DGH do not discharge until review by specialist centre)

Alloisoleucine Positive?

Original NBS card
Alloisoleucine Positive?

MSUD UNLIKELY
Presumptive false positive. Investigate and treat
Exclude liver disease including galactosaemia.

Possible intermittent MSUD
Send fibroblasts for enzymology. Manage as MSUD until result known.

MSUD CONFIRMED
Clinical management via specialist centre
Arrange sibling screening*****

* MSUD screening protocol see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

** MSUD initial clinical management guidelines www.bimdg.org.uk

*** MSUD diagnostic protocol for confirmatory tests see Laboratory Guide for IMDs https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

**** MSUD Dietetic Management Pathway www.bimdg.org.uk/site/guidelines-enbs.asp

***** MSUD sibling testing see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot
IVA Clinical Referral Pathway

*IVA Suspected

**ON THE SAME DAY**
1. NSL inform Specialist team who will inform Designated team, depending on baby’s location, to arrange urgent hospital admission.
2. Specialist/Designated team to CONTACT FAMILY
3. Specialist/Designated team to instruct family to go to appropriate hospital with 24 hr paediatric cover. Offer to arrange an ambulance
4. If initial review not at Specialist Centre, Specialist team to liaise with the hospital (Designated or on call Paediatric Consultant)
   a. Fax/email information to the hospital for clinicians and parents, IVA A&E letter, ‘IVA is suspected’ leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the IMD specialist team.
   b. Clinical assessment by local paediatric team - admission to hospital regardless of clinical status
      i. Hospital to inform specialist centre when baby arrives & liaise with Specialist centre regarding clinical state if not already transferred.
      ii. Transfer to Specialist Centre as soon as appropriate
5. Commence clinical management** (Specialist or Designated team)
   a. Obtain diagnostic samples*** and send urgently to screening laboratory (by courier) inform lab
   b. Well baby – Ensure adequate feeding
   c. Unwell baby
      i. Obtain blood gases, U&E, LFT, FBC, cultures, urine ketone dipstick, site IV cannula
      ii. IV 10% dextrose infusion
      iii. Carnitine, specialist centre to organise supply and send to local hospital if necessary.
      iv. Reintroduce natural protein within 24-48 hours (refer to IVA dietetic management pathway****)
6. NSL inform GP, send IVA GP letter via fax / email copy to HV
7. NSL inform Local Screening Midwife by phone

**FIRST REVIEW within 24 hours of screening result**
If not in specialist centre, speak directly via telephone or other communication
Review available test results. Continue to follow dietetic management pathway as required****
(If at DGH do not discharge until agreed by specialist centre)

Specialist team to review face to face within 2 working days. Arrange to feedback remaining results by 5 working days

- Plasma C5>cutoff
- Urine IVG normal
- Urine MBG normal
- Urine PVA increased
- Effect of antibiotics (PIVAA (Pivalate))
- IVA unlikely
  - No treatment needed - discharge
- Plasma C5<cutoff
- Urine IVG normal
- Urine MBG normal
- Urine PVA normal
- False positive
- Short/branched chain acyl-CoA dehydrogenase deficiency (SBCAD).
  - Long term FU. No ER
- Plasma C5>cutoff
- Urine IVG normal
- Urine MBG normal
- Urine PVA normal
- Uncertain Repeat investigations
- Plasma C5>cutoff
- Urine IVG normal
- Urine MBG normal
- Urine PVA increased
- IVA CONFDIRMED
  - Arrange sibling screening*****
  - PTO
IVA Clinical Referral Pathway (continued)

** T allele detected

- Feed normally.
- Glucose polymer emergency regimen for intercurrent illness.
- See dietary management guidelines ****
- Weaning/post-weaning FU visits and others as needed.

** T allele not detected

- Start/continue with carnitine (100mg/kg/day in 2-4 divided doses) and/or glycine (150mg/kg/day divided in 3 doses) supplementation.
- If clinically unwell consider protein restriction (See dietary guidelines****
- Emergency regimen

---

* IVA screening protocol see Laboratory Guide for IMDs for details
www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

** IVA initial clinical referral guidelines and standards www.bimdg.org.uk

*** IVA diagnostic protocol for confirmatory tests (Laboratory Guides for IMDs)
https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

**** IVA dietetic management pathway for details see
www.bimdg.org.uk/site/guidelines-snbs.asp

***** See IVA sibling testing - Laboratory Guide for IMDs for details
www.gov.uk/topic/population-screening-programmes/newborn-blood-spot
GA1 Clinical Referral Pathway

* GA1 Suspected

ON THE SAME DAY
1. NSL inform Specialist team who will inform Designated team if required (depends on babies address) to arrange urgent hospital admission
2. Specialist/Designated team CONTACT FAMILY
3. Specialist /Designated team to instruct family to go to appropriate hospital with 24 hr paediatric cover. Offer to arrange an ambulance.
4. If initial review not at Specialist Centre, Specialist team to liaise with the hospital (Designated or on call Paediatric Consultant)
   a. Fax/email information to the hospital for clinicians and parents, ‘GA1 is suspected’ leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the Specialist team.
   b. Clinical assessment & admission to hospital regardless of clinical status
      i. Hospital to inform specialist centre when baby arrives & feedback to specialist centre within 2 hours of admission if not already transferred to specialist centre.
      ii. Transfer to Specialist centre as soon as appropriate
5. Commence clinical management** (Specialist or Designated team)
   a. Consent for DNA testing to be obtained.
   b. Take diagnostic samples*** and send urgently to screening laboratory (courier) & inform lab
   c. Ensure adequate feeding
   d. 10% Dextrose infusion + carnitine if unwell. See GA1 dietetic management pathway****
6. NSL inform GP, send GA1 GP letter via fax / email & copy to HV
7. NSL Informs Local Screening Midwife by phone

FIRST REVIEW: FACE-TO-FACE - within 24 hours of screening result
Review diagnostic test results
(If at DGH do not discharge until review by Specialist centre)

Within 5 working days of diagnostic samples
Review available results and communicate with family

Biochemistry abnormal

Yes

1st FOLLOW-UP VISIT - within 5 working days of the 1st face-to-face review

No

1st FOLLOW-UP VISIT – with DNA results within 15 working days of the 1st face-to-face review

* GA1 screening protocol see Laboratory Guide for IMDs for details
** GA1 initial clinical management guidelines
*** GA1 diagnostic protocol for confirmatory tests
**** GA1 Dietetic Management Pathway
***** GA1 sibling testing
HCU Clinical Referral Pathway

** HCU suspected

** ON THE SAME DAY

1. NSL inform Specialist team who will inform Designated team depending on babies address. First review appointment arranged (to take place within 24hrs unless result available on a friday).
2. Specialist or Designated team CONTACT FAMILY
3. Specialist team to instruct family to go to specialist centre - if not possible go to appropriate hospital.
4. If initial review not at Specialist Centre, Specialist team to liaise with the hospital Designated Paediatrician (On-call Paediatric Consultant if not available) for assessment and treatment as indicate.
   a. Fax/email information to the hospital for clinicians and parents, ‘HCU is suspected’ leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the HCU specialist team.
   b. Hospital to inform specialist centre when baby arrives & feedback to specialist centre with a review within 2 hours of admission if not already transferred to specialist centre.
   c. Appointment with Specialist team arranged (within 24 hours)
5. Commence clinical management** (Specialist or Designated team)
   a. Take diagnostic samples*** inform screening lab including transport arrangements
   b. Provide supply of pyridoxine 50mg bd & folic acid (5mg/day) to be started if diagnostic tests confirm positive for HCU.
6. NSL inform GP, send HCU GP letter via fax / email & copy to HV
7. NSL informs Local Screening Midwife by phone

** WITHIN 5 working days of diagnostic samples

Review diagnostic results available and communicate results with family

HCU CONFIRMED

HCU UNLIKELY

Explain results to the family and discharge

Start pyridoxine 100mg per day & folic acid 5mg/day

Repeat plasma amino acids and total homocysteine after taking pyridoxine & folic acid for approximately 1 week and again after approx. 2 weeks.

(Results within 2 working days)

Discuss dietary treatment and provide family with methionine free formula. See HCU Dietetic management pathway****

Pyridoxine responsive (homocysteine in target range)

Yes

Communicate with family. Continue pyridoxine and folic acid

No/partial response

Arrange sibling testing*****

Communicate with family. Start methionine restricted diet - refer to HCU dietetic guidelines.

** HCU screening protocol see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

** HCU initial clinical management guidelines www.bimdg.org.uk

*** HCU diagnostic protocol for confirmatory tests see Laboratory Guide for IMDs https://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot

**** HCU Dietetic Management Pathway www.bimdg.org.uk/site/guidelines-enbs.asp

***** GA1 sibling testing see Laboratory Guide for IMDs for details www.gov.uk/topic/population-screening-programmes/newborn-blood-spot
6  Training and education

Training resources for the screening programme can be accessed via the following links:
- cpd.screening.nhs.uk/bloodspot-elearning
- cpd.screening.nhs.uk/interactivecard.php
- cpd.screening.nhs.uk/newbornbloodspot

Other useful resources can be found on the UK Screening Website [www.gov.uk/topic/population-screening-programmes/newborn-blood-spot](http://www.gov.uk/topic/population-screening-programmes/newborn-blood-spot)

A local policy for education and training for reflects National Guidance and should include the following:

<table>
<thead>
<tr>
<th>Policy</th>
<th>Local arrangements</th>
<th>Professional responsible</th>
</tr>
</thead>
</table>
| 1. Provision of an education and training programme for all healthcare professionals involved in the screening process, where appropriate utilizing tools and media provided by the UKNSC. | All newly employed healthcare professionals involved in the screening process will have training in the Trust’s newborn blood spot screening programme as part of their induction programme.
All healthcare professionals involved in the screening process will undertake an annual update session. | * [e.g. Local screening coordinator /practice development midwife, HV, lead paediatrician, dietitian].
Update training sessions to be organized by *.
All healthcare professionals responsible for ensuring annual update undertaken. |
| 2. All education and training provided should be evaluated and audited, where appropriate utilising tools and media provided by the UKNSC. | A record of attendance of training will be maintained.
All training sessions will be evaluated using the UKNSC audit tool. | *[e.g. Local screening coordinator /practice development midwife, HV, lead paediatrician, dietitian]. |
| 3. Annual review of training needs. | Review of training needs will be undertaken using the UKNSC audit tool. | * [e.g. Local screening coordinator /practice development midwife, HV, lead paediatrician, dietitian]. |
## 7. Quality Management and Clinical Governance

<table>
<thead>
<tr>
<th>Policy</th>
<th>Rationale</th>
<th>Local arrangements</th>
<th>Professional responsible</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. The clinical governance group [insert local name] is responsible for arranging release of audit data to identified regional and national screening teams in line with Caldicott and Governance arrangements.</td>
<td>To monitor compliance and adherence to national standards and assist in future strategic planning and commissioning of screening services.</td>
<td>[insert local arrangements].</td>
<td>Local Clinical Steering Group.</td>
</tr>
<tr>
<td>2. Provision of appropriate information for inclusion in national or local registers/audits must be provided.</td>
<td>To assist in the monitoring of the outcomes of screening and incidence of IMD.</td>
<td>[insert local arrangements].</td>
<td>*[name of identified lead].</td>
</tr>
<tr>
<td>3. The clinical steering group [insert local name] is responsible for ensuring appropriate links are in place with the Regional Newborn Blood spot Screening Operational</td>
<td>To enable effective performance management and quality assurance of locally delivered screening programmes.</td>
<td>[Insert names of groups and links and attendance requirements].</td>
<td>*</td>
</tr>
<tr>
<td>Policy</td>
<td>Rationale</td>
<td>Local arrangements</td>
<td>Professional responsible</td>
</tr>
<tr>
<td>----------------------------------------------------------------------</td>
<td>---------------------------------------------------------------------------</td>
<td>------------------------------------------------------------------</td>
<td>--------------------------</td>
</tr>
<tr>
<td>Management Group and input to/review the output from the Annual Regional Newborn Screening Governance Group</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4. All stakeholders (Acute Trust, CCG, CHRD, PHE) must contribute to the newborn screening annual audit pertaining to the previous fiscal year</td>
<td>All stakeholders have access to information regarding the screening programme.</td>
<td>[insert local arrangements for production of report].</td>
<td></td>
</tr>
</tbody>
</table>
### Regional contacts

<table>
<thead>
<tr>
<th>Team</th>
<th>Name</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Screening and Immunisation Lead, Public Health England,</td>
<td>Dr Alison MacKenzie</td>
<td><a href="mailto:alison.mackenzie6@nhs.net">alison.mackenzie6@nhs.net</a></td>
</tr>
<tr>
<td>Screening and Immunisation Manager, Public Health England,</td>
<td>Mr Matthew Dominey</td>
<td><a href="mailto:matthew.dominey@nhs.net">matthew.dominey@nhs.net</a></td>
</tr>
<tr>
<td>Director of Bristol Newborn Screening Laboratory</td>
<td>Dr Helena Kemp</td>
<td><a href="mailto:Helena.Kemp@nbt.nhs.uk">Helena.Kemp@nbt.nhs.uk</a></td>
</tr>
<tr>
<td>Deputy Director of Newborn Screening Laboratory</td>
<td>Ms Anny Brown</td>
<td><a href="mailto:Ann.Brown@nbt.nhs.uk">Ann.Brown@nbt.nhs.uk</a></td>
</tr>
<tr>
<td><strong>IMD Specialist Team:</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Consultant Metabolic Paediatricians</td>
<td>Dr Germaine Pierre</td>
<td><a href="mailto:Germaine.Pierre@UHBristol.nhs.uk">Germaine.Pierre@UHBristol.nhs.uk</a></td>
</tr>
<tr>
<td></td>
<td>Dr Effie Chronopoulou</td>
<td><a href="mailto:Efstathia.Chronopoulou@UHBristol.nhs.uk">Efstathia.Chronopoulou@UHBristol.nhs.uk</a></td>
</tr>
<tr>
<td>Regional Metabolic Dietitian</td>
<td>Camille Newby</td>
<td><a href="mailto:Camille.Newby@UHBristol.nhs.uk">Camille.Newby@UHBristol.nhs.uk</a></td>
</tr>
<tr>
<td>Regional Metabolic Specialist Nurse</td>
<td>Suzanne Cross</td>
<td><a href="mailto:Suzanne.Cross@UHBristol.nhs.uk">Suzanne.Cross@UHBristol.nhs.uk</a></td>
</tr>
<tr>
<td>Regional Quality Assurance Team ANNB (South West) Public Health England</td>
<td>Siobhan O’Callaghan</td>
<td>so’<a href="mailto:callaghan@nhs.net">callaghan@nhs.net</a></td>
</tr>
<tr>
<td>Senior Quality Assurance Manager</td>
<td>Wendy Ring</td>
<td><a href="mailto:wendyring@nhs.net">wendyring@nhs.net</a></td>
</tr>
<tr>
<td>Quality Assurance Managers</td>
<td>Maggie Denholm</td>
<td><a href="mailto:maggie.denholm@nhs.net">maggie.denholm@nhs.net</a></td>
</tr>
<tr>
<td>Regional Biochemical Genetics Diagnostic Laboratory</td>
<td>Dr Helena Kemp</td>
<td><a href="mailto:Helena.Kemp@nbt.nhs.uk">Helena.Kemp@nbt.nhs.uk</a></td>
</tr>
<tr>
<td><strong>CLIMB</strong></td>
<td></td>
<td><a href="http://www.climb.org.uk">www.climb.org.uk</a></td>
</tr>
</tbody>
</table>
9 Local contacts [insert local team]

<table>
<thead>
<tr>
<th>Team</th>
<th>Name</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Local Public Health Lead</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local Screening Coordinator</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local designated MCADD Specialist Team:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lead Paediatrician</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lead Dietitian</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local HV Lead</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local Biochemistry Laboratory Lead</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Local Child Health Records Department contact</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>