# ­NHS Newborn Blood Spot Screening Programme

Notification of inherited metabolic disease (IMD) diagnosis for babies/children not identified through the newborn blood spot screening

**Do not include any patient identifiable information on this form.**

Please send the completed form to: [england.annb-qa-admin@nhs.net](mailto:england.annb-qa-admin@nhs.net)

**Please attach a colour photograph of the blood spot card (front and back) if screening took place. This is to help assess whether blood spot quality may have been an issue.**

**Details of form completion**

|  |  |
| --- | --- |
| Form completed by (name) |  |
| Form completed by (job title) |  |
| Form completed by (email address) |  |
| Date form completed |  |

**Details of form sign off**

|  |  |
| --- | --- |
| Signed off by (name) |  |
| Signed off by (job title) |  |
| Signed off by (email address) |  |

**Details of the laboratory and baby/child detected***– do not include any patient identifiable data*

|  |  |
| --- | --- |
| Name of Newborn Screening Laboratory |  |
| Name of Lead Metabolic Clinician |  |
| Gestational age at birth |  |
| Year of birth |  |
| Ethnicity of baby (ONS code) |  |
| Date of detection |  |
| Details of clinical detection / presentation of affected patient |  |
| Age at initial clinical referral |  |
| Age at first appointment with designated clinician |  |
| Reason for referral to Metabolic Team |  |

**Section 1. IMD detected**

Please complete the diagnostic results for the condition identified.

**Section 1a – diagnostic results for PKU**

|  |  |
| --- | --- |
| Diagnostic Phe and Tyr results  (µmol/L and age at sample) |  |
| Pterins and DHPR results completed? | Yes / No  *If yes, give age and results* |
| Consistent with classic PKU/classic hyperphenylalanaemia? | Yes / No |
| Consistent with biopterin metabolism defect? | Yes / No  *If yes, detail clinical significance* |
| Consistent with variant/atypical hyperphenylalanaemia? | Yes / No  *If yes, detail clinical significance* |
| Please detail further investigations undertaken outside of PKU Diagnostic Protocol |  |

**Section 1b – diagnostic results for MCADD**

|  |  |
| --- | --- |
| Blood acylcarnitines  (µmol/L and age at sample) |  |
| c.985A>G mutation analysis | Homozygous / Heterozygous / None |
| Qualitative urinary organic acids completed? | Yes / No  *If yes, give results* |
| Organic acids consistent with MCADD? | Yes / No |
| Quantitative urinary hexanoylglycine completed? | Yes / No  *If yes, give age and result (µmol/mmol creat)* |
| Extended mutation screening (EMS) complete? | Yes / No  *If yes, give age at result, EMS laboratory, result and any commentary on clinical significance as reported from EMS laboratory* |
| Was there a previous diagnosis of:   * Not MCADD * MCADD unlikely * Likely unaffected MCADD carrier * Unaffected MCADD carrier? | *If yes, please specify* |
| If yes to the above, please detail further biochemical investigations undertaken outside of MCADD Diagnostic Protocol |  |

**Section 1c – diagnostic results for MSUD**

|  |  |
| --- | --- |
| Blood amino acids (alloisoleucine)  (µmol/L and age at sample) |  |
| Qualitative urinary organic acids completed? | Yes / No  *If yes, give results* |
| Please detail further investigations undertaken outside of MSUD Diagnostic Protocol |  |

**Section 1d – diagnostic results for IVA**

|  |  |
| --- | --- |
| Blood acylcarnitines  (µmol/L and age at sample) |  |
| Qualitative urinary organic acids completed? | Yes / No  *If yes, give results* |
| c.941C>T (932C>T) mutation analysis | Homozygous / Heterozygous / None |
| Quantitative urinary isovalerylglycine completed? | Yes / No  *If yes, give age and result (µmol/mmol creat)* |
| Please detail further investigations undertaken outside of IVA Diagnostic Protocol |  |

**Section 1e – diagnostic results for GA1**

|  |  |
| --- | --- |
| Blood acylcarnitines  (µmol/L and age at sample) |  |
| Qualitative urinary organic acids completed? | Yes / No  *If yes, give results* |
| Full gene sequencing | Homozygous / Heterozygous / None |
| Please detail further investigations undertaken outside of GA1 Diagnostic Protocol |  |

**Section 1f – diagnostic results for HCU**

|  |  |
| --- | --- |
| Blood amino acids (methionine and total homocysteine)  (µmol/L and age at sample) |  |
| Qualitative urinary organic acids completed? | Yes / No  *If yes, give results* |
| Liver function test, folate, vitamin B12 |  |
| Please detail further investigations undertaken outside of HCU Diagnostic Protocol |  |

**Section 1g – diagnostic results for HT1**

|  |  |
| --- | --- |
| Bloodspot or plasma succinylacetone |  |
| Qualitative urinary organic acids completed? | Yes/ No  *If yes, give results* |
| Blood amino acids (tyrosine) (µmol/L and age at sample) |  |
| *FAH* gene sequencing | *Homozygous / Heterozygous / None* |
| Please detail further investigations undertaken outside of HT1 Diagnostic Protocol |  |

**Section 2. Screening results**

**Was the baby screened?** If yes, complete section 2a, 2b and 2c.

If no, go to section 3.

**Section 2a**

|  |  |
| --- | --- |
| Clinical diagnosis before screening | Yes / No |

**Section 2b**

|  |  |
| --- | --- |
| Age at first blood spot |  |
| Was the sample sent via post or courier? |  |
| Age at receipt of sample in laboratory |  |

**Section 2c – screening result for the condition the baby has been identified with**

|  |  |  |
| --- | --- | --- |
| **PKU** | Screening, Phe = | Comments / observation / other information |
| **MCADD** | Screening, C8 = | Comments / observation / other information |
| **MSUD** | Screening, xLeu = | Comments / observation / other information |
| **IVA** | Screening, C5 = | Comments / observation / other information |
| **GA1** | Screening, C5-DC = | Comments / observation / other information |
| **HCU** | Screening, Met = | Comments / observation / other information |
| **HT1** | Screening, SUAC = | Comments / observation / other information |

**Section 3. Reason**

**Section 3a – not screened (not eligible)**

|  |  |
| --- | --- |
| Infant / child moved into UK over 1 year of age |  |
| Baby died prior to screening | *Please indicate hours / days of age* |

**Section 3b – not screened (eligible)**

|  |  |
| --- | --- |
| Parents declined screening for IMD |  |
| Parents declined further investigations |  |

**Section 3c – false negative screen**

|  |  |
| --- | --- |
| Baby / child who was screened and reported as ‘condition not suspected’ that subsequently has a confirmed diagnosis and the screening result is confirmed |  |
| Had any of the following occurred:   * dextrose administration prior to blood collection * later screening of a mover-in or ‘older’ baby * delayed transit of sample (>14 days) * blood transfusion <72 hours prior to blood collection * contaminated card with no repeat sample available | *If yes, please specify* |

**Section 3d – screening programme deviation**

|  |  |
| --- | --- |
| No sample taken |  |
| Sample not taken at appropriate time |  |
| Sample did not arrive in laboratory |  |
| Test failure / lab error:   1. NBS lab 2. Genetic lab | *Please specify* |
| Screen positive baby not referred / did not attend clinical appointment (no failsafe) |  |
| Other cause | *Please specify* |

**Section 4. Investigation**

**Section 4a – blood spot card**

|  |  |
| --- | --- |
| Expiry date |  |
| Condition of card |  |
| Retrospective review of blood spot quality |  |
| Retrieval and re-analysis of original blood spot sample (and samples either side) | *If sufficient sample, confirm analysis on TMS and/or a more specific method e.g. IEC or LCMSMS* |

**Section 4b – laboratory**

|  |  |
| --- | --- |
| Detailed review of laboratory data from the time of sample, to include:   * IQC performance * EQA performance * in-house population data * bias relative to national population monitoring performance * accreditation status at the time that the screening lab was analysed |  |

**Section 4c – clinical history and results**

|  |  |
| --- | --- |
| Any relevant neonatal clinical history at the time of blood spot sampling, such as antibiotics or dextrose | *If yes, please specify* |
| Information missing from maternal history  (e.g. the use of antibiotics) | *If yes, please specify* |
| Investigation results after clinical presentation, to include blood and urine metabolites, and genetics |  |

**Section 4c – siblings** *(only complete if applicable)*

|  |  |
| --- | --- |
| Maternal phe or other primary marker |  |
| Genetic results of index case and affected case |  |

**Section 4d – findings summary**

|  |  |
| --- | --- |
| Root cause |  |
| Contributory factors |  |
| Possible implications for patient / programme |  |

**Section 4e – further investigation**

|  |  |
| --- | --- |
| \*Has SQAS been informed? | Yes / No |
| \*Has this been reported as an incident? [www.gov.uk/government/publications/managing-safety-incidents-in-nhs-screening-programmes](http://www.gov.uk/government/publications/managing-safety-incidents-in-nhs-screening-programmes) | Yes / No |

*\*NB – This is not automatically needed, it is well known that not all cases of homocystinuria, GA1 or MSUD will be detected by screening and there may be adequate reasons to explain a false negative result for other disorders. Each case should be assessed on an individual basis to determine if a possible error contributed to a false negative result.*

**Section 4f – additional comments**

|  |  |
| --- | --- |
| Please include any other additional comments that may be relevant. |  |

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