MCN for Neonatology

West of Scotland

Neonatal Guideline

Newborn Blood Spot Screening

Introduction

This guideline is applicable to all medical, nursing and midwifery staff caring for the newborn in the West of Scotland. It has been written to comply with the recommendations laid out in the QIS (Quality Improvement Scotland) Clinical Standards Document[1] published in October 2005 and updated in July 2008[2]. These standards aim to ensure that Newborn Blood spot screening is offered to **all** babies. The standards also aim to ensure that the collection of samples, laboratory testing and initiation of treatment in positive cases are completed efficiently so that affected babies achieve maximum benefit from early and appropriate treatment.
Staff should also familiarise themselves with the contents of the updated information leaflets for parents:

- **"You're pregnant"**
- **"Your Baby"**

The Newborn Screening Programme

From 23rd March 2026, the Scottish Newborn blood spot screening programme will include eleven conditions:

1. Phenylketonuria (PKU)

Autosomal recessive

Incidence in Scotland approximately 1 in 8,000

Test detects elevated levels of phenylalanine

Early treatment prevents the development of serious, permanent learning disability

Screen positive babies are reviewed immediately: the diagnosis should be confirmed and treatment with a low phenylalanine diet started by 21 days of life

2. **Congenital hypothyroidism (CHT)**

Incidence 1 in 3,500

Test detects raised levels of TSH (thus ***will not*** detect rare cases of hypothalamic hypothyroidism)

Early treatment essential to prevent or minimise learning disability

Screen positive babies are reviewed immediately at the neonatal unit, thyroid function checked and investigations ordered after discussion with the on-call endocrinologist Thyroid replacement therapy is usually started at this visit or the following day, and arrangements must be made (by telephone) for prompt follow up at the Royal Hospital for Children, Glasgow.

3. **Cystic Fibrosis (CF)**

Autosomal recessive

Incidence 1 in 2,500

Test detects raised levels of immunoreactive trypsin: confirmation is by DNA testing from the same blood spot

Cases referred to respiratory team, and for genetic counselling. Babies should be seen and started on treatment by day 35

Screening will identify some carriers who will be offered an appointment with a genetic counsellor (this is dealt with in general practice)

4. **Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)**

Autosomal recessive

Incidence 1 in 10,000 to 1 in 20,000

Impaired fat metabolism places the child at risk of severe hypoglycaemia, particularly with intercurrent illness. First presentation may be sudden death

Average age at presentation in unscreened populations is 14 months. Mortality is 15-20% and up to 30% of survivors sustain brain damage

Management is to prevent metabolic crises by ensuring an adequate supply of carbohydrate

5. **Sickle Cell Disease (SCD)**

Autosomal recessive

Incidence 1 in 2500, mostly in Black African children

Carries a risk of sickle crises which may be complicated by infections, anaemia, stroke and/or death

Affected babies should receive pneumococcal vaccine and prophylactic penicillin

Testing may detect carriers: they will be offered an appointment with a genetic counsellor

6. Maple syrup urine disease (MSUD)

Autosomal recessive

Incidence of ~1 in 180,000

Amino acid metabolism defect, resulting in excess leucine, isoleucine and valine.

Usually manifests in the first couple of weeks with failure to thrive, feeding difficulties, lethargy, irritability and/or a maple syrup odour in the urine and cerumen. Untreated, it can rapidly lead to irreversible neurological damage, seizures, encephalopathy and death.

There are even rarer milder/intermittent variants, the blood spot screening only picks up some of these.

7. Isovaleric acidaemia, (IVA)

Autosomal recessive.

Incidence of ~1 in 155,000

Amino acid metabolism defect, with deficient or absent isovaleryl-CoA dehydrogenase enzyme, resulting in excess isovaleric acid and a failure to breakdown leucine.

This usually presents in the neonatal period with vomiting, seizures, poor feeding, hypotonia, ketoacidosis, quickly progressing to coma and death. Less commonly it can present with a more chronic intermittent type through childhood with failure to thrive, developmental delay and learning disability. A characteristic "sweaty feet" odour can often be identified during episodes of acut metabolic decompensation.

Diagnostic confirmation is with urinary organic acid analysis.

Management is with a low-protein, leucine-restricted diet, use of carnitine and/or glycine to reduce isovaleric acid levels, and specialist metabolic clinician and dietetic support.

8. Glutaric aciduria type 1 (GA1)

Autosomal recessive.

Incidence of ~1 in 100,000.

Amino acid metabolism defect, with deficient or absent glutaryl-CoA dehydrogenase, resulting in an inability to sufficiently break down lysine and tryptophan and results in excess glutaric acid in the blood.

Usually presents in the first 1-2years with an infection-related metabolic decompensation, with hypoglycaemia, hyperammonaemia and encephalopathy; can also present with childhood subdural haemorrhage or intracranial thrombosis, developmental delay or macrocephaly.

Diagnosis is confirmed with urine organic acid and plasma acylcarnitine analysis.

Management is with a low-protein, low-tryptophan/lysine diet, especially in the first 6years, carnitine supplementation, avoidance of fasting and specialist metabolic dietician and clinician input.

9. Homocystinuria (HCU)

Autosomal recessive inheritance.
Incidence of ~1 in 200,000.
Amino acid metabolism defect, with failure to breakdown methionine to cysteine, resulting in excess homocysteine and methionine.
Can present in infancy with slow failure to thrive and developmental delay, and in older children with visual symptoms, eye pain and/or headache, skeletal and connective tissue anomalies, decreased bone density, and thrombosis.
Diagnostic confirmation is with urine and blood for raised homocysteine and methionine levels.
Management is with pyridoxine (helps convert homocysteine to cysteine), betaine (helps break down homocysteine), a low-protein diet, specialist metabolic clinician and dietetic support.

10. Hereditary tyrosinaemia Type 1 (HT1) AKA Hepatorenal tyrosinaemia.

Rare, autosomal recessive

Incidence around 1 in 100,000

This is a protein metabolism problem. A missing enzyme - fumarylacetoacetate hydratase - results in an inability to break down the amino acid tyrosine. As this builds up, so does the levels of succinylacetone (SUAC), which is harmful and can be measured in the blood.

This places the baby at risk of failure to gain weight, jaundice, diarrhoea, vomiting, fever, hepatomegaly, and spontaneous bleeding and bruising.

Unscreened infants usually present within the first 6 months with evolving acute liver failure or renal dysfunction. Older children can occasionally present with the above, or with hepatocellular carcinoma, acute painful extremity or abdominal crises, hypertension, hyponatraemia, and/or respiratory failure/death.

Untreated, children die before aged 10, but an early diagnosis combined with consistent adherence to dietary protein restriction and use of Nitisinone (inhibits catabolism of tyrosine) can result in 90% reaching adulthood.

Screen positive individuals are reviewed same day in their local hospital, and blood (including genetic) and urine testing would be undertaken, and a provisional treatment plan started alongside discussion with metabolic medicine specialists.

11. **Spinal Muscular Atrophy (SMA)**

Spinal muscular atrophy has been added as part of an in-service evaluation (ISE) for a **period of 18-24months**, with screening for SMN1 and SMN2 genes, and therefore **may be only temporarily available**.

Incidence of around 1 in 10-15,000.

This is a neuromuscular condition causing progressive muscle wasting and weakness, including muscles involved in breathing and swallowing. There are 4 main types in infants/children (a 5th type, SMA 0, has an onset pre-natally and accounts for only 2% of cases), type 1 being the most common and earliest to present. This can be with recurrent aspiration and pneumonia and delayed motor milestones alongside the muscle weakness and wasting.

***As one of the potential therapies for SMA is steroids, we would recommend delaying administration of live vaccines (particularly BCG) until after SMA status is known. This avoids any possible delays in treatment.**

Blood spot specimen analysis is done in a specific way, which affects consent:

- Sickle cell disease, Cystic fibrosis, Congenital Hypothyroidism and Spinal Muscular Atrophy are tested individually so **consent can be for any combination of these.**
- PKU, MSUD, IVA, GA1, Homocysteinuria, MCADD and HT1 are tested together, and so **consent can be for ALL of these or NONE of these**.

Informing the Parents about the test

All Parents/Carers should receive clear information (written or in other formats) to help them to make an informed choice about newborn screening.

- Outline information about newborn blood spot screening is available in the "Ready Steady Baby" book which is given to all mothers-to-be at booking.
- Parents should be directed to the information resource links on Page 1 of this document. If language is an issue then an interpreter should be arranged to ensure that an **informed** decision can be made. Individual arrangements must also be made for parents and/or carers with visual or learning difficulties which prevent them from reading the material.
- Information about the test should have been received at least 48hours before the blood spot specimen is taken and parents/carers must be given the opportunity to ask questions of an appropriately trained health professional, using an interpreter if required. *If the questions asked by the parents/carers cannot be answered by the health professional then they should be referred to a senior member of the medical staff*.

Consent

- Written consent must be taken before the sample is acquired and the completed form retained in the Health Record. The consent form should be completed and retained even if testing is declined. ***See Appendix for Consent form***
- Consent will usually be taken from the mother. If this is not possible then consent may be sought from another person with parental responsibility. If no person with parental rights is available to give consent then advice should be sought from the Central Legal Office.
- If one or more of the screening tests is declined parents/carers should be informed of the action they should take if they change their minds or if symptoms or signs of the condition appear in their child. Signs and symptoms may be non-specific, and so any change in the child's condition should result in prompt review by either the midwife or GP. Parents should also be informed that some or all of the benefits of screening may be lost if the test is delayed. Note that parents must accept all decline all 7 metabolic conditions; it is no longer possible to opt out of some. Parents may opt out of all 7 metabolic conditions, but still accept other screening tests.
- This information is available in written form (*See Appendix*) but staff must still ensure that the parents/carers understand the information. A standard letter should be sent to the General Practitioner informing them that the infant has not been tested when this is the case. (*See Appendix*).
- If parents change their mind shortly after the screening test was offered then the blood spot screen should be performed. A subsequent change of mind should be dealt with by the GP or Health Visitor who can liaise with the screening laboratory or local Paediatrician to determine the most appropriate means of testing.
- **If the infant is premature, generally unwell, or shows specific signs or symptoms of one or more of the conditions covered by the newborn screening programme then testing for these conditions should proceed in the child's best interests.** Hopefully the parents will be persuaded of the clinical need for these investigations but failing this, advice should be sought from the Central Legal Office.

Timing of the test

Blood spot screening tests should be offered to all babies between 96 and 168 hours of life. 95% of positive CHT and PKU cases should have started treatment by 14 days of age, unless deliberately delayed for further testing.

- The blood spot specimen should be obtained on day 5 of life wherever possible *including bank holidays and weekends*. The date of birth is counted as day 0, regardless of the time of birth (NB this is at variance with the Badger information system which counts date of birth as day 1).

Local Arrangements to ensure that a blood spot specimen is obtained prior to a red cell transfusion

- The timing of the first specimen should not be delayed, even if the baby is premature, unwell or parenterally fed. Repeat specimens are required for premature infants - s*ee below.*
- The specimen should be mailed to the screening laboratory the same day. *When parents decline the screen the unfilled blood spot card must still be returned to the screening lab with all the patient information complete.*
- A repeat specimen may be required where the initial specimen is inadequate or where the result was equivocal. Repeat sampling will be performed by midwifery/neonatal nursing staff irrespective of the age of the child. Repeat samples must be taken and mailed to the laboratory within 24 hours of notification by the laboratory.

Repeat testing for sick and premature infants.

A protocol should be in place for screening babies who are ill, transfused or born prematurely. Repeat testing will be required in the following circumstances

- **Transfused Babies** If the baby has received a transfusion before the 5 day bloodspot the result of thyroid and cystic fibrosis screening may be unreliable so the laboratory will request a repeat sample, 72 hours after the last transfusion. Note that this is in addition to the pre-transfusion sample required to exclude Sickle Cell Disease.
- **Premature Babies**
  If the baby was very premature (<32 weeks)
  A further sample should be taken for retesting for hypothyroidism at 28 days of life

NB – *it is no longer necessary to repeat a sample from a baby who was not enterally fed at the time of the 5th day sample.*

Babies with affected siblings, or whose parents are known carriers of one of the screened conditions

Where a previous sibling is affected by one of these disorders, or the parental carrier status is known to put this infant at increased risk the baby may need to be managed differently.

Sickle Cell Disease (SCD)

Where both parents are Sickle Cell disease carriers, there is a 25% risk of the child being affected. This is of no clinical consequence in the neonatal period and so there is no clinical need for testing before the routine day 5 test. The latter should be carried out as normal, with a note on the test card regarding the family status to aid the screening laboratory.

In circumstances where there are particularly high levels of parental concern, consideration can be given to taking a blood spot for screening earlier (this must not be cord blood), but this is not encouraged. Testing should be done only after discussion with a consultant, and it must be made clear that the routine day 5 test is still required to screen for other disorders. The card should be clearly marked as an early sample, and sent to the newborn blood spot screening lab, with the laboratory called in advance to discuss. Samples should not be sent to local haematology laboratories for analysis. A clear plan for feedback of results to the family must be made.

Positive results obtained through the routine (day 5) screening process will be dealt with by the screening laboratory with a direct referral to the paediatric haematology services, or to genetic counselling services where carrier status is identified.

Cystic Fibrosis

Where both parents are carriers of Cystic Fibrosis this should be discussed antenatally and a plan made. The plan should be clearly documented in the maternal notes.

In most circumstances cord blood is taken at birth and genetic testing for the parental CFTR mutations performed. For this to be successful, knowledge of the parental mutations is required, which should be dealt with antenatally if early neonatal testing is planned. If in doubt cord bloods can be taken, sent to the DNA lab in clinical genetics and stored pending discussion with the family and the attending consultant geneticist.

Congenital Hypothyroidism

This is rarely an inherited disorder so no specific measures need be taken unless specified antenatally. This should be specified on the paediatric section of the yellow alert sheet of the maternal notes.

Spinal Muscular Atrophy

Types 1-4 are caused by a mutation at the SMN1 gene on Chromosome 5. Inheritance of this is autosomal recessive. These families will usually have had involvement with genetics prenatally, and any plan around testing at birth should be actively sought and followed. In the absence of this, assessment of the infant should proceed promptly and discussion with the neuromuscular team (if symptomatic) and clinical genetics (if asymptomatic) should be early and judicious.

All Inherited Disorders of Metabolism
(PKU, MCCAD, MSUD, IVA, GA1, HCU, HT1)

This varies according to the condition, and the **most optimal and safest way for this to happen is with prompt engagement and advance planning with the metabolic team early in the obstetric journey**.
The metabolic team are keen to ensure individualised plans are in place for these siblings/family members, and will often be aware of them already, but there is also helpful and regularly updated national guidance on the BIMDG website including for the at risk sibling, linked here: Prospective management of at risk sibling – BIMDG

To make contact with the metabolic team for such siblings on a **non-urgent** basis (i.e. during pregnancy), liaise with the metabolic consultant already involved directly, or email: **ggc.paediatricmetabolic.clinicalenquiries@nhs.scot**

To do this on an **urgent** basis (i.e. after birth where no clear individualised plan is evident), make contact with the metabolic consultant directly through Glasgow RHC switchboard 0141 201 0000 (in hours/out of hours), or the direct number 0141 452 4406 (in hours).

Individual plans, to include necessary testing and fluid/feed management, made at any stage should be clearly documented and easily available as a "neonatal alert" or similar in the obstetric notes and should be shared with colleagues as necessary to ensure prompt and appropriate care.

Heelprick sampling

Method

1. Wash hands and put on gloves
2. Check baby's name and age with the parents (*or identity bands if in hospital*)
3. Expose baby's heel
4. If cleansing is required allow to dry completely before sampling
5. Do not use Vaseline on the heel
6. Hold foot firmly, encircling heel with thumb and index finger
7. With one movement pierce the soft tissue of the outer aspect of the heel with the safety lancet
8. Release pressure and allow blood to flow
9. Completely fill all circles on blood spot card with blood ensuring that the blood soaks right through the card. Fill the circles from the centre. Try to avoid contact between the absorbent paper and the baby's heel or your gloves (note that samples where blood has been applied inappropriately or where the spots have been compressed/layered will be rejected – see below)
10. Allow the blood spots to air dry completely before placing in the glassine sleeve. Only one card per glassine sleeve
11. Using the cotton wool ball, apply slight pressure to the puncture site to stop the bleeding
12. Apply spot plaster if required; this should be removed during the next episode of care
13. Dress baby and leave comfortable
14. Dispose of safety lancet in sharps box
15. Wash hands
16. Ensure the card is placed in a FREEPOST envelope and posted the same day

The Scottish Screening laboratory has employed enhanced criteria for accepting blood spot samples, bringing the Scottish Screening laboratory into line with evidence based agreed UK national criteria for sample quality.

Why does the blood spot quality matter?

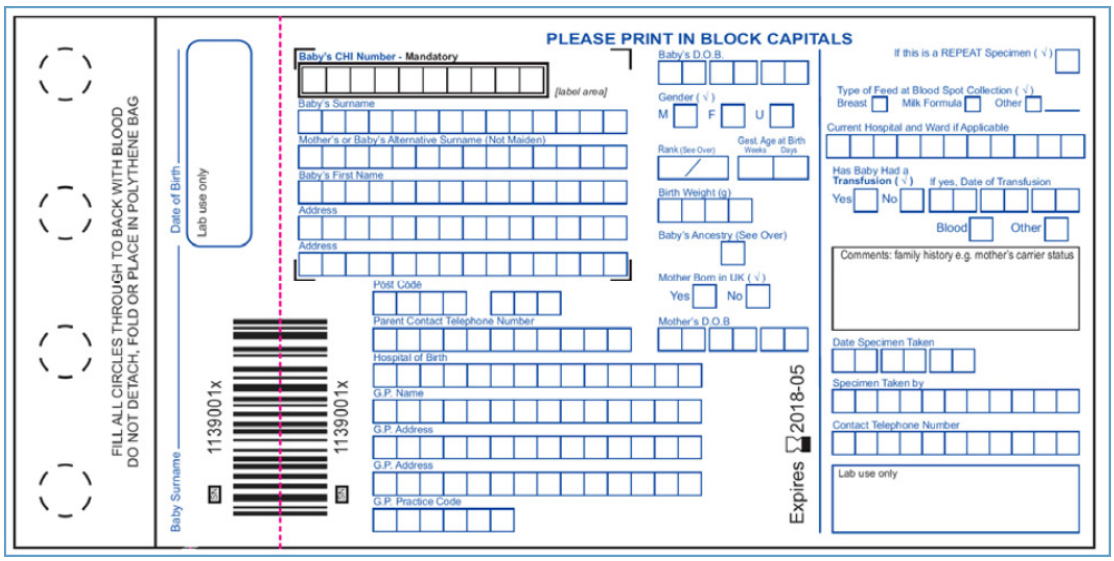
- Good quality blood spot screening samples are vital for ensuring that babies with rare but serious conditions are identified and treated early. Poor quality samples can result in delayed referral and diagnosis of affected babies.

The most significant effects of poor quality samples are:

- Falsely low analyte concentrations (false negative results), which can be caused by:
  - Small volume spots (i.e. under-filled circles)
  - Compression of the sample

  - Falsely high analyte concentrations (false positive results), which can be caused by:
    - Layering of blood samples
    - Applying the blood to the front and the back of the card

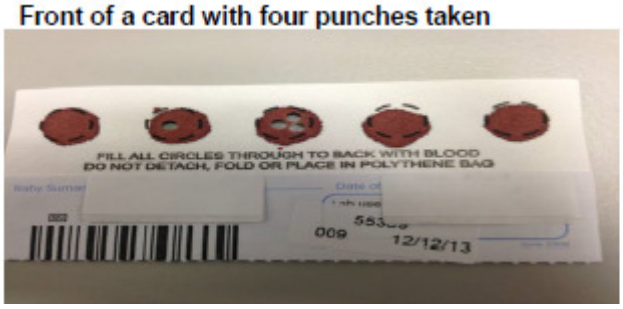
Please remember that it is extremely important that the blood fills the circles and soaks through to the back of the card. If the bloodspots are too small or not soaked through to the back of the card it is very likely that the sample will have to be repeated.



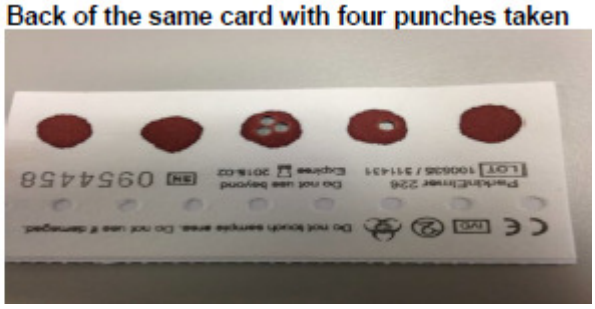
Examples of samples- good and bad (NB – photos from old 5 spot screening cards)

Good quality samples:

Front of a card with four punches taken



Back of the same card with four punches taken



Insufficient samples:

Front of card

Back

Blood not soaked through to the back of the card:

Front of card

Back

Layered sample

This is where one spot of blood is layered directly on top of another, or the blood has been applied to one side of the card and then turned over and reapplied to the other side.

Front

Back

Sample placed in glassine envelope before it had completely dried:

Documentation

- **The Blood spot card** - Accurate completion of the data requested on the blood spot card cannot be over emphasised. Too much time is wasted in checking incorrect or missing information. Ensure all details on the blood spot card are completed (*especially the CHI number*) and sign the card. If the baby fulfils any of the criteria for repeat screening (*see above*) record this in the comments box on the blood spot card along with other important clinical information (*eg* delayed passage of meconium or echogenic bowel on antenatal scan).
- **Computerised Discharge Report / Discharge documentation** – A copy of the postnatal discharge documentation is sent to the GP on discharge from the postnatal ward. This will contain details of the blood spot test if performed before discharge
- **Scottish Handheld Maternity Record (SWHMR) -** Record heelprick sampling (or refusal of the test) in the postnatal record within the SWHMR.
- **Neonatal Case Records -** Babies who are in the neonatal unit at the time of testing must have a record of the blood spot test(s) in the medical/nursing notes according to local policy. This will include an entry in the Badger electronic record in units where this is in use. If the baby fulfils any of the criteria for repeat testing a plan for a repeat testing should be carefully documented.
- **Transfer letter / Badger Discharge summary –** If the baby is transferred to another hospital written documentation regarding newborn screening should form part of the transfer documentation

Additional Local Arrangements for documentation

Informing the Parents

All Parents/carers should be informed of the timescale within which the results will be made available and the format in which they will be communicated and by whom.

95% of results should be issued from the laboratory to an appropriate health care professional within 2 working days of the receipt of the specimen by the laboratory

All positive results should be communicated to parents/carers as soon as possible after the screen and no later than 14 days from the specimen collection for PKU and CHT and 27 days from specimen collection for CF

Currently there are no arrangements to communicate all screening results to the parents/carers. They should be told that the test results are available within 2 working days of receipt of the sample by the screening laboratory, and that they will be contacted directly if the screen indicates that repeat testing or further investigation is required. This contact would usually occur within 1-2 days of the screen result being obtained by the laboratory. It is important to ensure that accurate contact telephone details are obtained from parents at the time of testing.

Who has parental rights for consenting?

The child's father may have parental rights and responsibilities if:-

(1) he is married to the mother
(2) he is named as the father on the birth certificate
(3) he has been assigned parental rights by a court order
(4) the mother has sought a 'parental responsibilities and parental rights agreement'.

Parental Responsibilities and Parental Rights Agreement (Scotland) Amended Regulations 2006.

Another person may have parental responsibilities if they have been appointed legal guardian by court order. However most other carers, whether they are relatives of the mother or foster carers assigned by social work, will not have parental rights. If a baby is going to be cared for by a person without parental responsibility consent should be obtained from the mother prior to discharge from the maternity unit.

Information Leaflets

Newborn Blood Spot Screening Information for Healthcare Professionals - 2017

Blood spot Screening Consent Form – See Appendix 1

Parental Information leaflet to be issued when screening declined – See Appendix 2

Letter to GP informing them that screening has been declined – See Appendix 3

Links

1. **Pregnancy and newborn screening standards – Healthcare Improvement Scotland**
2. **Pregnancy and Newborn Screening Developments Website**
3. **Newborn screening tests | NHS inform**

Appendix 1

Consent for Newborn Blood Spot Screening

Consent for Newborn Blood Spot Screening for Congenital Hypothyroidism (CHT), Cystic Fibrosis (CF), Sickle Cell Disorders (SCD), Spinal muscular atrophy (SMA)*, and 7 Inherited Metabolic Disorders (IMDs) - *Phenylketonuria (PKU), Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), Maple Syrup Urine Disease (MSUD,* Homocystinuria (HCU), *Isovaleric Acidaemia (IVA),Glutaric aciduria type 1 (GA1) and Hereditary Tyrosinaemia Type 1 (HT1).*

I have received and read the national patient information leaflet and have had an opportunity to discuss the tests I am being offered with a health professional. I understand the reasons for the tests and the consequences of the results. I also understand the significance of not having these tests performed. I am aware that my decision whether or not to have these tests will not affect the quality of care delivered by healthcare professionals.

Baby's Name……. Date of birth …/…/…

CHI Number……

I wish **I do not wish** my baby to be tested for Congenital Hypothyroidism

I wish **I do not wish** my baby to be tested for Spinal Muscular Atrophy*

I wish **I do not wish** my baby to be tested for Cystic Fibrosis

I wish **I do not wish** my baby to be tested for Sickle Cell Disorders

I wish **I do not wish** my baby to be tested for the 7 IMDs

I understand that the Newborn Blood Spot Screening Programme needs to obtain my permission to store the newborn blood spot card beyond the initial 12 month testing period and to use any of the blood spots left over after testing is complete for anonymised research, such as the development of new screening tests. If any research was proposed in which the researcher would be able to me or my baby, a member of staff from the Newborn Bloodspot Screening Programme would always contact me again to seek my approval.

I agree **I do not agree** for the storage of my baby's blood spot sample beyond the initial 12month testing period.

I agree **I do not agree** to the use of any leftover blood spots for anonymised research.

* SMA testing is part of an 18-24month in-service evaluation (ISE), beginning March 2026, and testing as part of this screening programme may therefore be temporary.

Signature:…… (Parent/Guardian) Date …/…/…

Signature:……(Witness: Healthcare professional)

Print:……

Designation: ……. Date …/…/…

Appendix 2 – Patient Information Leaflet

Information for parents when the bloodspot test has not been undertaken

The aim of the newborn bloodspot screening programme is to identify specific conditions, as soon after birth as possible and before the onset of clinical symptoms

Newborn bloodspot screening can identify the following conditions:

- Phenylketonuria (PKU)
- Congenital Hypothyroidism (CHT)
- Cystic Fibrosis (CF)
- Sickle Cell Disorders (SCD)
- Spinal muscular atrophy (SMA)*
- Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)
- Glutaric Aciduria type 1 (GA1)
- Homocystinuria (HCU)
- Isovaleric Acidaemia (IVA)
- Maple Syrup Urine Disease (MSUD)
- Hereditary Tyrosinaemia Type 1 (HT1)

Screening allows these conditions to be detected early – usually before signs and symptoms become apparent. Early treatment can help prevent brain damage, serious illness, or in some cases death, in the case of PKU, CHT, GA1, HCU, IVA, MSUD, HT1 and MCADD. Early treatment can also limit the effects of CF, SCD and SMA*. Delays in diagnosis may be detrimental to your child's health.

Because your baby has not been screened you should look out for any of the symptoms listed below. If you are concerned, please contact your GP for advice. Let them know that your baby has not been screened for one or more of the above conditions. By the time you notice anything is wrong it may be too late to prevent some lifelong damage but treatment, if started as early as possible, may still be of some benefit

If you wish your baby to be screened for any or all of the conditions please contact your GP or Health Visitor immediately. Testing is best carried out as early as possible. Screening becomes less reliable for CF after the baby is six weeks old although other tests can be used to diagnose CF at a later stage.

Symptoms and signs of Congenital Hypothyroidism (CHT)

- Constipation
- Dry skin
- Hoarse cry
- Large tongue
- Swelling around the eyes
- Feeding problems
- Sleeping for long periods
- Prolonged jaundice
- Baby fails to thrive and develop

Signs and Symptoms of Cystic Fibrosis (CF)

- Recurrent Infections of the lungs and sinuses
- Chesty cough that doesn't clear up
- Dirty nappies smell much worse than normal and are pale and greasy
- Poor growth and poor weight gain
- Salty taste to the skin

Signs and Symptoms of Sickle Cell Disorder (SCD)

- Increased susceptibility to infection
- Painful swelling of the fingers and hands or toes and feet. Pain can also occur in the arms, legs, back and abdomen (from about 6 months)
- Pale skin and lips
- Yellow pigment in the eyes and skin (jaundice)

Signs and Symptoms of Phenylketonuria (PKU)

- Baby fails to progress in learning to sit, use the hands, crawl and develop speech
- Unusual behaviour, such as screaming episodes, repetitive rocking, head banging or arm biting
- Vomiting leading to weight loss
- Irritability / crying a lot
- Skin problems, such as sensitivity to light, dry skin or itchy rashes
- Baby grows and develops more slowly than normal
- An unusual smell to the skin, hair and urine
- Fits / seizures

Phenylketonuria after childhood

There is a rare mild form of PKU, which causes little obvious effect, apart from mild learning difficulties. Boys and girls can reach adulthood and be unaware that they have this form of PKU. However if your child is a girl it is particularly important that she is aware that she has not been tested for PKU. If your daughter does have PKU, this may harm the development of any children she may have. She needs to know that she has not been tested for PKU to allow her to make choices about her future pregnancies. She may choose to be tested for PKU. Children born to boys with PKU are not affected in this way.

Signs and symptoms of Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)

Signs and symptoms can develop quickly in infants who are not feeding well or who are unwell with an infection or illness such as diarrhoea and vomiting

- Drowsiness
- Seizures
- Coma

Signs and symptoms of Glutaric Aciduria type 1 (GA1)

In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Early signs may be:

- Vomiting
- Irritability
- Excessive sleepiness
- Floppiness
- Breathing difficulties

Signs and Symptoms of Homocystinuria (HCU)

- Learning difficulties
- Eye problems
- Bones which are abnormally long and thin (osteoporosis)
- Blood clots / strokes

Signs and Symptoms of Isovaleric Acidaemia (IVA)

Children with IVA can become severely unwell. Early signs may be:

- Vomiting
- Excessive sleepiness
- Floppiness
- Rapid breathing

Signs and Symptoms of Maple Syrup Urine Disease (MSUD)

Many babies with MSUD become unwell when they are a few days old, with:

- poor feeding
- vomiting
- excessive sleepiness
- Coma or permanent brain damage

In older children a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. The early signs in older children include confusion and poor balance as well as loss of appetite, vomiting and excessive sleepiness. As with babies, this can lead to a coma unless treated correctly

Signs and Symptoms of Hereditary Tyrosinaemia Type 1 (HT1)

Babies with this condition can suffer with:

- failure to gain weight or thrive
- vomiting and diarrhoea
- jaundice
- abdominal swelling caused by an enlarged liver
- unexplained bleeding or bruising

Older children may present with any of the above, or with raised blood pressure, electrolyte imbalances, severe breathing difficulties, kidney disease or liver failure.

Signs and symptoms of Spinal Muscular Atrophy (SMA)*

Babies with this condition will struggle to use their muscles well.
They may have:

- difficulty swallowing and frequent episodes of choking
- frequent chest infections related to milk getting into the lungs
- not meeting milestones with gross motor skills (rolling/sitting/crawling etc)
- wasting of the muscles
- arms and legs appear weak and “floppy”

This gets progressively worse with time.

* SMA testing is part of an 18-24month in-service evaluation (ISE), beginning March 2026, and testing as part of this screening programme may therefore be temporary.

Appendix 3

Neonatal Bloodspot Screening Test Declined

Dear Doctor,

This letter is to inform you that a patient registered with your practice has declined the offer of neonatal bloodspot screening for their baby, for one or more of the screened conditions. They have been issued with an information sheet, a copy of which is enclosed, detailing the symptoms, signs and long term effects of these conditions. The leaflet makes clear that late detection of these conditions can leave the child with irreversible sequelae. The leaflet also indicates that they should seek urgent medical advice should they change their minds or if they believe that their child is developing the symptoms of one of the screened conditions. If such concerns are raised with yourself please refer the child to the local paediatric unit without delay.

Mother's Details

Name …… DOB……

Unit Number …… CHI Number ……

Address ……..

Date of Delivery…….

Name of Baby……

Baby's CHI Number ……..

Screening Test(s) Declined:

Congenital Hypothyroidism ☐

(Phenylketonuria, MCADD, Maple syrup urine disease,
Isovaleric acidaemia,
Glutaric aciduria type 1, Hereditary tyrosinaemia type 1
& Homocystinuria)

NB These conditions must be accepted or declined together ☐

Cystic Fibrosis ☐

Sickle Cell Disorders ☐

Spinal Muscular Atrophy* ☐

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Update March 2026

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