



Newborn Bloodspot Sampling Guidelines

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V1.0

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Introduction

Wherever possible these Guidelines are based with kind permission on the English Newborn Bloodspot Screening Programme Bloodspot Taking Guidelines. The aim of the Newborn Blood Spot Screening Programme is to identify specific conditions, as soon after birth as possible and before the onset of recognisable clinical symptoms.

The 6 Inherited metabolic disorders Phenylketonuria (PKU), Congenital Hypothyroidism (CHT), Cystic Fibrosis (CF), Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), Sickle Cell Disorders (SCD), Maple Syrup Urine Disease (MSUD), glutaric aciduria type 1 (GA1), isovaleric acidaemia (IVA) and homocystinuria (HCU) are offered as a package so screening is either for all of the conditions or none of them. Additionally, all babies moving into Scotland from elsewhere who have not been screened/have a verifiable screening result should also be offered screening up to 1 year of age. Parents may decline any or all of the tests (the 6 IMD conditions count as one test) and must be informed that their baby remains eligible for screening within the Programme up to the age of 1 year.

For the small number of babies affected, early detection, referral and treatment can help to improve their health and prevent severe neurological impairment or even death. Without early treatment, the conditions screened for can result in:

PKU	permanent neurological impairment and profound additional needs
CHT	permanent, serious physical problems and additional learning needs
CF	poor weight gain, frequent chest infections and reduced life expectancy (symptoms can be present even with treatment)
MCADD	serious illness and possible death
SCD	severe pain, life-threatening infections and anaemia (symptoms can be present even with treatment)
MSUD	coma, permanent neurological impairment and possible death
GA1	coma and neurological impairment
IVA	coma, permanent neurological impairment and possible death
HCU	Additional learning needs, eye problems, osteoporosis, blood clots or strokes

These guidelines aim to:

- Provide a consistent and clear approach to offering newborn blood spot screening and taking the sample
- Provide a framework for Health Professionals to support parents in making informed choice about newborn bloodspot screening for their baby
- Support Health professionals in obtaining good quality samples to prevent the need for avoidable repeats
- Reduce pain and discomfort during the heel puncture

Why is the quality of the bloodspot so important?

Good quality blood spot samples are vital to ensure that babies with rare but serious conditions are identified and treated early. Poor quality samples can cause inaccurate newborn screening results and therefore these samples cannot be accepted by the laboratory. The most significant effects of poor quality samples are:

- 1) Falsely low analyte concentrations (false-negative results), which can be caused by:

- Small volume spots (i.e. under-filled circles)
 - Compression of the sample
- 2) Falsely high analyte concentrations (false-positive results), which can be caused by:
- Layering the blood
 - Applying the blood to the front and the back of the card

Poor quality blood spots could therefore lead to false-negative and false-positive screening results – **this means that babies with a condition might be missed or referred for further tests unnecessarily**. If poor quality blood spots are received, the newborn screening laboratory will have to request a repeat sample. Avoidable repeat samples can cause anxiety for parents, distress to babies and delays in the screening process (for example a baby might miss CF screening because it can only be screened for accurately up to eight weeks of age). They are also a waste of healthcare resources. In some cases, parents may refuse to consent to a repeat – this means that the baby will have incomplete screening and an opportunity to identify a condition in an effective timeframe to prevent irreversible serious health consequences may be missed.

The Scottish Newborn Screening Laboratory (SNSL) have adopted the UK, evidence based consensus on blood spot quality with standardised acceptance and rejection criteria. To ensure that an avoidable repeat sample is not requested, sample takers are advised to obtain four good quality blood spots and complete all the fields on the blood spot card accurately (**always check the expiry date on the card before use**).

Good quality blood spots are those where the circle is filled and evenly saturated by a single drop of blood that soaks through to the back of the blood spot card.

3. Offer of screening

Parents should be asked at booking if they have a family history of any of the inherited metabolic diseases (IMDs) so that early newborn screening can be offered and arranged if appropriate.

It is important that parents can make an informed choice about screening for their baby and are prepared for the blood sampling procedure. Ideally parents should be advised of the screening options offered for their baby and provided with a copy of the national screening leaflet to support them to make an informed decision at least 48 hours prior to taking the blood sample. The information leaflet will provide information on the conditions screened for, how the bloodspot will be taken and how the results will be received. Where applicable provide the leaflet in the appropriate language, some translated versions and also alternative formats are available for download from Public Health Scotland and others can be arranged by contacting them. If the required language is not available, alternative arrangements should be made with local interpreter services.

It is the responsibility of the NHS Board through the Screening Coordinator and the Director of Public Health, to ensure that robust systems are in place locally to confirm that every newborn baby or child up to the age of 1-year-old, resident in their NHS Board is invited to participate in screening. Midwives working in hospital or in the community are responsible for ensuring that testing is offered and national protocols are followed which includes, in Scotland, the requirement is for written consent. If the baby is still in hospital when the baby is between 96-120 hours of life (4-5 completed days)-, it is the responsibility of the ward, Neonatal Intensive Care Unit (NICU), Special Care Baby Unit (SCBU), or the Surgical Unit staff to obtain the blood spot sample.

2. Parental consent and refusal

Parents should have a pre-test discussion with the healthcare professional taking care of them and their baby regarding the newborn blood spot test. Discussions should include the purpose, processes and benefits of the test. Parents will be asked to complete a consent form stating that they have received sufficient information to understand the reasons for testing, the significance of the results and the possible consequences of not having tests performed. When obtaining consent for the newborn blood spot screening programme, you must ensure that parents understand they are consenting to the following:

- The sample being taken
- The sample being booked in and analysed in the newborn screening laboratory and used for quality assurance
- The laboratory sending the results to the child health record department
- The results being stored on the child health information system
- The potential identification of their baby as a “carrier” of SCD or CF
- A referral to specialist services if a result is positive
- The bloodspot card being stored for a minimum period by the laboratory to allow for retesting if required and then to be stored indefinitely unless permission for ongoing storage was declined
- Their baby’s anonymised data being used for research studies that help to improve the health of babies and their families in the UK, for example population studies, unless they specifically decline this.
- In rare circumstances, receipt of invitations from researchers who would like to use their baby’s blood spot card for named research

The consent form should be completed accepting or declining each individual test (the 6 inherited metabolic disorders (PKU, MCADD, MSUD, GA1, IVA and HCU) are offered as a package), storage of the card and the use of samples for anonymised research. This information should also be recorded on the bloodspot card in the comments section. The form should be countersigned by the healthcare professional taking the blood sample and filed in the maternity records. The midwife will also ascertain family contact details for the first month and establish the preferred method of communication, should it be necessary to repeat testing.

If the parents have declined screening for one or more of the conditions, the midwife must still complete the blood spot card in the usual manner, recording each condition declined and the reason (if stated) in the comment section. Parents should be made aware they have the option to take up screening at a later date up until the baby is one, with the exception of screening for Cystic Fibrosis as the screen can only be offered up until they are 8 weeks of age, and who they should contact. They should also be informed sheet **“When the blood spot screening test has not been undertaken”**. The information sheet lists the possible signs and symptoms of the conditions that are screened for. This information is also passed on to the child health records department and reported to the family’s GP and Health Visitor, so that the GP/Health Visitor can also look out for these signs and symptoms.

3. The newborn bloodspot card

It is vital that every section of the blood spot card is completed accurately. The information supplied is entered onto the Scottish Newborn Screening Laboratory’s (SNSL) Laboratory Information Management System (LIMS) and is used for:

- the identification of the infant – to ensure that the correct result is issued to the correct baby

- the determination of results – certain parameters have to be fulfilled before results are valid and issued
- ensuring the correct protocol is followed – different protocols are applied in particular tests
- There are two fields which act as a check to ensure that the bloodspot has been taken within the correct timescales; time of birth and time specimen taken.

4. Completing the bloodspot card

Check the expiry date before completing the blood spot card. **Expired cards should not be used, they should be returned to the SNSL.**

The middle section of the front of the card contains demographic details

It is best practice to use labels for this section if available. Always ensure that it is the baby's label and not mother's.

CHI number	The use of the CHI number is a mandatory requirement and should always be noted in full. Ensure the baby's CHI number is supplied and not the mothers.
Mother's or Baby's Alternative Surname	If more than one name is known please document both
Address	This is the home address of the baby at the time of birth
Post code	This should be in full
Parent contact telephone number	This is important as immediate contact with parents may be required
Hospital of birth	Name of hospital or if born before arrival then the hospital that the baby is delivered in. Home should be stated if not associated with a hospital

GP	General Practitioner with whom the baby will be registered
GP address	Name of surgery and address
Baby's DOB and time of birth	This must be accurate as the interpretation of results for some tests is dependent on the age of the child. If the sample is taken too early, i.e. before 4 completed days of age, this can invalidate the results
Gender	Tick box; M-Male F-Female U-Unknown or Uncertain
Rank	This is important to indicate whether baby is a twin etc as shown on the back of the card eg 1/1, 1/2. This will avoid the specimen being treated as a duplicate specimen of another baby
Gestation	This is the gestation that the baby was born at and is important for some of the tests. Mark in weeks and days
Baby's Ancestry	Choose from one of the family origin codes, as detailed on the back of the card
Mother born in the UK	This is used in relation to some epidemiological studies
Mothers DOB	Used as another link to the baby if details differ from those held by Child Health Information System

The right hand side of the front of the card is information used by the laboratory:

Type of feed	If more than one type applies tick both i.e. breast and bottle. 'Other' is for total parenteral nutrition (TPN) in cases of sick babies.
Current Hospital and ward if applicable	If the baby is still in hospital please supply the hospital and ward, this will allow child health to locate the baby if a repeat sample is needed.
Has baby had a transfusion	Date of transfusion must be included. Please also indicate if the transfusion was a blood transfusion or other product such as platelets or FFP. The interpretation of results will differ depending on whether blood or other products were given.
Comments	This space should be used to record any condition(s) where screening has been declined (specify which conditions and see guide for additional steps when screening is declined). In addition, it should be used to record 'No Storage' or 'No Research' if parents have requested this. This space should also be used to record: <ul style="list-style-type: none"> • any relevant antenatal screening results • parent(s) known carriers of haemoglobin variants/CF, • Meconium Ileus • any family history relevant to the conditions screened for • nipple cream used by mother, as this can result in false positive isovaleric acidaemia (IVA) • reason for sample if not taken between 96-120 hours (e.g. pre transfusion, preterm CHT)

	<ul style="list-style-type: none"> • mover in baby • congenital hypothyroidism (CHT) preterm day 28/discharged home sample • repeat sample for inconclusive cystic fibrosis (CF) • repeat sample for borderline CHT
Date and time Specimen Taken	This is essential. The SNSL uses this to determine age of baby when sample was taken. Cards without this date will not be issued with a result until this has been established. The time the specimen has been taken must also be provided to allow the laboratory to confirm that the baby is over 96 hours old
Specimen Taken By	Name of healthcare professional
Contact Telephone Number of Health Professional	Will allow immediate contact in the event that the laboratory or clinical service needs to make direct contact with the healthcare professional
Lab use only	Must be left blank. This will be used for the laboratory's unique numbering system

The left hand side of the front of the card is for the specimen collection

- **Baby's Surname** and **DOB** must also be completed on this portion of the card as it shall be separated from the rest of the card and this is required for identification purposes in addition to the barcode included on both sections of the card.

The reverse side of the card describes the following:

- **Family origins** – This information is important for SCD testing
- **Rank** – This gives birth order in multiple pregnancies
- **Blood Collection** – Brief instructions

<p>Baby's Ancestry</p> <table border="0"> <tr><td>Code</td><td>Description</td></tr> <tr><td>A.</td><td>African or African-Caribbean (Black)</td></tr> <tr><td>B.</td><td>South Asian (Asian) e.g. India</td></tr> <tr><td>C.</td><td>South East Asian e.g. China</td></tr> <tr><td>D.</td><td>Other Non-European (Other) e.g. North Africa</td></tr> <tr><td>E.</td><td>Southern & Other European (White) e.g. Greece, Poland</td></tr> <tr><td>F.</td><td>United Kingdom (White)</td></tr> <tr><td>G.</td><td>Northern European (White) e.g. Austria</td></tr> <tr><td colspan="2"><i>Any other European family origins e.g. Australia, S. Africa, N. America</i></td></tr> <tr><td>H.</td><td>Don't Know</td></tr> <tr><td>I.</td><td>Declined to Answer</td></tr> <tr><td>J.</td><td>Any Mixed Background</td></tr> </table>	Code	Description	A.	African or African-Caribbean (Black)	B.	South Asian (Asian) e.g. India	C.	South East Asian e.g. China	D.	Other Non-European (Other) e.g. North Africa	E.	Southern & Other European (White) e.g. Greece, Poland	F.	United Kingdom (White)	G.	Northern European (White) e.g. Austria	<i>Any other European family origins e.g. Australia, S. Africa, N. America</i>		H.	Don't Know	I.	Declined to Answer	J.	Any Mixed Background	<p>Rank</p> <p>Identifies birth order: singleton, twins, triplets</p> <table border="0"> <tr><td>1/1</td><td>Singleton</td></tr> <tr><td>1/2</td><td>Twin 1</td></tr> <tr><td>2/2</td><td>Twin 2</td></tr> <tr><td>1/3</td><td>Triplet 1 Etc.</td></tr> </table>	1/1	Singleton	1/2	Twin 1	2/2	Twin 2	1/3	Triplet 1 Etc.
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<p>EC REP Wallac Oy, Mäntymäki 6,  PerkinElmer, P.O. Box 100, Turku, Finland, FIN-20521, Finland, 2018-05-28 2021-05-28</p> <p>Date of Production: 03-2015</p>																																	
<p>CE LVD  Do not touch sample area. Do not use if damaged.</p> <p>PerkinElmer 226 Do not use beyond LOT 103649 / 315416 Expires 2018-05 SN 1139001</p>																																	

5. Timing of the sample

Ideally samples are taken between 96-120 hours of life (4-5 completed days), day of birth is day 0, and sent on the day of sampling to the Scottish newborn screening laboratory. In exceptional circumstances samples can be sent between day 5 and day 8. Completing the time, the sample was taken, as well as the date is necessary to allow the laboratory to confirm that the baby was at least 96 hours old. Eligible 'movers in' should be offered screening as soon as possible.

Additional tests are offered to babies born preterm and babies at risk of blood transfusion and if required by a screening protocol to achieve a conclusive result. Babies who are premature, unwell, or have had blood transfusions should all have the newborn blood sample taken as usual and this information must be recorded on the blood spot card in the comment box.

Records are kept of all tests including those declined. As noted before, even if all screens are declined a card should still be completed and sent to the laboratory indicating that all screening has been declined to ensure records are complete and the family is not contacted for a missed screen in error.

6. Preparation for taking the bloodspot sample

In order to take the newborn blood spot sample, the following is required:

- Blood spot card (check expiry date), glassine envelope and prepaid/addressed envelope
- Non-sterile protective gloves
- Age appropriate automated incision device designed for use on newborns
- Cotton wool/gauze
- Spot plaster (if required)
- Sharps box
- Maternity/baby record
- Water for cleansing

7. Collecting the bloodspot sample

Recommend comfort measures for the baby. Ensure the baby is cuddled and in a secure position for taking the sample, swaddling the baby may reduce pain/discomfort. Engaging the baby through face to face contact, voice and touch may be beneficial. Suggest that the baby is breast fed during the heel prick as an analgesic (or expressed breast milk). Whilst there is no evidence that formula feed has analgesic properties, parents may comfort formula-fed babies with a feed. Alternatively, a pacifier may provide comfort.

- Explain the procedure to parents. Complete all boxes on the card including the baby's surname and date of birth on the blood spot sample section to the left of the card. When completing the card care must be taken to place the card on a clean surface to avoid contaminating the sample.
- Ensure the baby is comfortable and in a secure position for taking the sample
- Clean the heel by washing thoroughly with tepid, plain water. The water should not be heated and the baby's foot should not be immersed. If faecal matter cannot be removed from the foot with water, use a mild, unperfumed soap to clean away the faecal matter and then rinse the foot thoroughly. This is because faecal matter

contains very high concentrations of immunoreactive trypsinogen (IRT). IRT is measured during screening for CF therefore faecal contamination may lead to a false positive result. Do not use alcohol or alcohol wipes.

- Wash hands and apply gloves. The heel should be warm (but in most cases additional prewarming of the foot is not required) and completely dry. Soft paraffin solutions should not be used as they can alter the result of the screen and can clog the equipment used.
- Obtain the sample using an automated incision device designed for use on newborns. An arch-shaped incision device is recommended. Skin puncture must be no deeper than 2.0 mm. The external and internal limits of the calcaneum are the preferred puncture sites (shaded areas diagram A).
- The whole heel plantar surface may be used in term and pre-term infants who have repeated heel punctures to a penetrative depth of no more than 1.00mm (shaded area diagram B). Avoid the posterior curvature of the heel. Allow the heel to hang down to assist blood flow. This reduces the soft tissue damage and pain from repeated heel puncture in the same area.
- Before activation place the automated incision device against the heel in accordance with manufacturer's instruction. This is to ensure the correct depth of incision is achieved – not too deep to cause harm to the baby, and not too shallow to prevent adequate blood flow.

Diagram A

For full and preterm infants



Adapted from Jain and Rutter

Diagram B

For infants who have had repeated heel punctures



The aim is to fill each 4 circles on the blood spot card using a single drop of blood. Wait for the blood to flow and a hanging drop to form. Allow one spot of blood to drop onto each of the circles on the blood spot card. Do not allow the heel to make contact with the card. Do not squeeze the foot in an attempt to increase blood flow. The first drop of blood should be used. Allow the blood to fill the circle by natural flow, and seep through from front to back of the blood spot card. Fill each of the circles completely. Spots that exceed the dotted lines on the filter paper are acceptable provided that a single drop of blood has been used. Do not compress or apply pressure to the blood spots (for example when sealing the postage envelope) as this reduces the density of blood on the sample and there is a significant risk that this could lead to a “suspected” result being missed.

If the blood flow ceases the congealed blood should be wiped away firmly with cotton wool or gauze, to disturb the clot and encourage blood flow. Gently massage the foot, avoid squeezing, and drop the blood onto the bloodspot card. If the baby is not bleeding a second puncture is necessary. The second puncture should be performed on a different part of the

foot, as marked by the shaded areas in diagrams A and B. When the sample collection is complete, wipe excess blood from the heel and apply gentle pressure to the wound with cotton wool or gauze. Apply a hypoallergenic spot plaster if required and remind the parent to remove the plaster in a few hours.

Ethylenediaminetetraacetic Acid (EDTA) is an anticoagulant used in some blood collection tubes. EDTA can interfere with the interpretation of testing and could lead to false negative results. Blood from EDTA tubes, citrate tubes or capillaries should never be used to fill blood spots as this will affect the measurement of Thyroid-Stimulating Hormone (TSH) for CHT and Immunoreactive Trypsinogen (IRT) for CF leading to false negative results.

8. Good quality samples

Initially the lab needs to take four 3.2mm ‘punches’ from the sample that is provided. If the result from one of the punches is above the cut-off for one of the conditions tested for, then the test has to be repeated to confirm the original result. To do this two further punches for each test which gave a high result are taken. This would mean that another two to eight punches may be required. Additionally, sometimes a further sample for mutation analysis (e.g. as in the protocol for cystic fibrosis screening), will require a further two punches. This means that up to 14 punches may be required. This is another reason good quality bloodspots are required.

Front of a card with four punches taken



Back of the same card with four punches taken



This is a very good quality sample: i.e. there is plenty of blood and it is soaked all the way through.

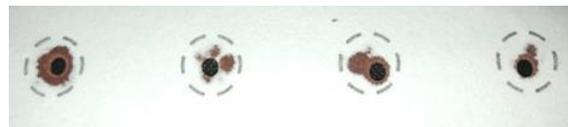
9. Insufficient samples

Research has shown that the levels of analytes tested for by newborn screening can be compromised when a small sample size is used. As a result, the minimum sample size the SNSL can accept is 8mm in diameter. This means that cards such as the following are insufficient and would be rejected resulting in an “avoidable repeat”.

Front of card (less than 8mm)



Back of the same card (less than 8 mm)



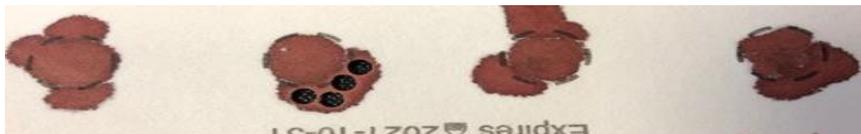
Samples will also be rejected as insufficient if there is not enough blood to perform all the tests required or if the blood has not soaked all the way through to the back of the card (i.e. white paper can be seen where there should be blood) then there is a risk of a false negative result as the concentration of the sample will be diluted and an affected baby could be missed. The following samples are insufficient, because the blood has not soaked through to the back of the card properly and what has come through is patchy with white paper visible on the punched spots.

Front of card

Back



Samples will also be rejected if it appears the incorrect application technique has been used to apply the blood to the sample. This includes layered samples, 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 re-applied on the other side, and multi-spotted samples, where the heel has been dabbed against the sample resulting in several small spots. The following is an example of layering, you can see where the original spot partially dried and then more blood was added, creating a 'two spot' effect.



Finally, samples will not be accepted if the blood it looks like the blood spot has been compressed. Compressed blood spots pose a significant risk to the baby as the levels can decrease by up to 25%. This can post a very significant risk of cause a false negative result, which means that a baby with a disease is not detected. This could be life threatening. Compressed samples are very difficult for the lab to spot but one way is when the is a significant amount of blood of the glassine, as in the picture below. Always ensure that the sample is completely dry before placing it in the glassine cover.



For further information on sample quality, please contact the Scottish Newborn Screening Laboratory on 0141 354 9277 or email GG-UHB.NewbornScreeningLaboratory@nhs.net

10. After taking the bloodspot sample

Check the completed bloodspot card with the parent and make any necessary changes and record that the sample has been taken in the maternity record, in compliance with local protocols. The blood spot card should be air dried flat in a horizontal position and then placed in a glassine envelope provided with the card and sent directly to the Scottish Newborn Screening Laboratory in the pre-addressed, free post envelope as soon as possible after collection, ideally on the same day and no more than 24 hours after taking the sample. Only one blood spot card should be placed in each glassine envelope. No more than six blood spot cards should be placed in a single envelope as this will exceed the contracted postal weight and delay the delivery of specimens. Dispatch should not be delayed in order to batch cards together for postage. If a post box is used, ensure it is one

that is emptied daily (Monday to Saturday). If the baby is in hospital, record and notify the baby's screening status on discharge/ transfer notifications.

In the event of a postal strike or severe weather which may disrupt deliveries, staff should refer to their local NHS Board protocols and contingency plans to ensure the timely delivery of blood spot cards to the SNSL.

Screening is offered routinely to all babies whose mothers are known or suspected to be infected with HIV or Hepatitis B. The blood spot card should be identified as a Biohazard. The envelope in which the card is placed must not be marked as 'Biohazard' to avoid breach of confidentiality.

11. Repeat samples

Occasionally it may be necessary to repeat a test and a second blood sample will need to be collected. If a repeat sample is required, the SNSL notifies the relevant Child Health/Screening Department to arrange for a repeat sample to be obtained by the midwife or health visitor.

There are a number of reasons why a repeat sample may be requested:

- There was insufficient blood available to perform all tests
- The blood spot card was damaged or did not reach the laboratory
- Layering or compression of the blood
- Equivocal or borderline test results – this means that the test result is not abnormal enough for the baby to be referred to a specialist but is not completely within normal ranges. There are several reasons why tests give an inconclusive result and often the repeat specimen provides a definitive result. If the repeat sample is also inconclusive then arrangements will be made for the baby to be assessed by a nominated clinical specialist
- There were insufficient details on the card to allow accurate analysis of the results
- The baby was too young when the specimen was collected (less than 96 hours old)
- The card was not dried properly prior to being posted
- The analysis was unsatisfactory due to specimen contamination or deterioration
- The specimen took more than 14 days to reach the laboratory and was therefore unsuitable for testing
- The baby was premature (less than 32 weeks' gestation at birth)
- The baby had a blood or blood product transfusion less than 72 hours before the sample was taken

The 'repeat sample' box should be ticked on the blood spot card.

12. Special circumstances

12.1 Babies born prematurely or cared for in neonatal units

Babies admitted to neonatal units are likely to have multiple bloodspot samples taken. Blood spot screening should be coordinated with other tests where possible to minimise the number of invasive procedures performed. Venepuncture or venous / arterial sampling from an existing line is an alternative method to collect the blood spot sample. This is providing the sample is not contaminated with EDTA/heparin and the line is cleared of infusate. Do not use heparinised capillary tubes as lithium heparin is difficult to detect and can affect DNA testing.

Babies less than 96 hours of age should have a single circle bloodspot sample taken on admission/prior to blood transfusion for the routine screening test for SCD. This should be on a separate bloodspot card marked "Pre-transfusion" and should be completed fully as described previously. Tape or a sticky label can be placed over the unused circles to avoid the routine sample being added to the pre-transfusion bloodspot card.

The '**Pre-transfusion**' blood spot card should be stored with the baby's medical records in line with local protocols and despatched to the newborn screening laboratory together with the routine sample if the baby has received a blood transfusion in the interim, to prevent the need for DNA analysis to complete SCD screening. The single circle blood spot sample taken and marked as '**Pre-transfusion**' can be discarded appropriately if the baby does not receive a blood transfusion.

If the baby is transferred to another unit before the routine sample has been taken, ensure the pre-transfusion blood spot card accompanies the infant. Details of newborn sampling should be documented and included in transfer information.

The routine blood spot sample (four spots) should be taken as soon after the baby reaches 96 hours old as possible for all babies regardless of medical condition, medication, milk feeding and prematurity.

When a baby has had a blood transfusion or blood product, either intrauterine or in the newborn period, an interval of at least 3 days (72 hours) is required between the transfusion and the routine blood spot sample for CF, CHT, MCADD and PKU to enable metabolite concentrations to return to pre-transfusion levels (for intrauterine transfusion count day of birth as date of transfusion). In the event of multiple blood transfusions however, or if a baby is on extra corporeal membrane oxygenation (ECMO), even if it has not been 3 days (72 hours) since the last transfusion, a routine blood spot sample should be sent by day 8 at the latest regardless to ensure all babies are screened by day 8 regardless of blood transfusion status.

In this scenario, a repeat sample will be needed at least 3 clear days after the last transfusion or the end of ECMO. The date of the last blood transfusion or end of ECMO must be recorded on the blood spot card and on discharge/transfer notifications.

If a baby has not had a pre-transfusion sample taken, the laboratory may forward the routine sample to the DNA laboratory for analysis as a failsafe however an additional cost would be incurred and not all conditions screened for by the SCD screen will be detected.

Provider organisations should ensure failsafe arrangements for notifying screening status when the care of babies is transferred. This includes babies who are transferred in the neonatal period. The screening status of the baby is to be recorded on an auditable system and included in the discharge/transfer documentation.

Babies born at less than 32 weeks (equal to or less than 31 weeks + 6 days) require a second blood spot sample to be taken in addition to the routine sample taken 96 hours after birth to ensure a valid sample for congenital hypothyroidism screening as immaturity can mask this condition. These babies are to be tested when they reach 28 days of age (counting day of birth as day 0) or day of discharge home, whichever is the sooner.

Complete the details on the blood spot card as described previously, recording 'CHT preterm' on the blood spot card so the laboratory is aware of the reason for the second sample. Write the gestational age on the card. If the baby is being discharged home before 28 days of age, write '**discharged home**' on the blood spot card, again this is so the

laboratory knows why the repeat sample was taken before day 28. The responsibility for taking each sample lies with the healthcare professional that is responsible for clinical care at the time the blood spot sample is due to ensure babies who are transferred at less than 28 days of age have all newborn blood spot tests completed.

Prematurity can mask CHT. Babies born at less than 32 weeks (less than or equal to 31 weeks + 6 days) require a second blood spot sample (two spots) to be taken, in addition to the day 5 sample. The explanation to be given to parents is that in babies born at less than 32 weeks of pregnancy, the routine test may not pick up congenital hypothyroidism. It is advised to have another test at either 28 days of age or immediately before the baby is discharged home, whichever comes first. Mark the card '**CHT preterm**'. Write the gestational age on the card.

If the baby is being discharged home before 28 days of age, write '**discharged home**' on the repeat sample. Record all blood spot samples taken in baby's hospital records, on transfer documentation, PCHR and on an auditable IT system

12.2 Babies transferring into area (up to the age of 1 year)

For a child transferring into the NHS Board from within the UK who is under 1 year of age, the Child Health/Screening Departments should check any available Health Visitor records received from the previous NHS Board of residence and document any results available for the child. If there are no records or results available, the previous NHS Board or health provider, if known, should be contacted to ascertain whether screening took place and if any written results are available. If a child under 1 year of age has transferred in from abroad and has no verifiable documented screening results available in English screening should be offered for all nine conditions.

If a child is under a year of age (up to but not including their first birthday) and has no documented results (or declines) then screening should be offered for all the untested conditions **only** if the blood spot sample can be taken no later than 14 calendar days after the baby's first birthday. Samples should not be taken later than 14 calendar days after the baby's first birthday as they will not be eligible for screening. If the process is to offer and take the sample on the same day, this can only be done before the baby's first birthday. Parents should be informed that the results may not be as reliable as when taken at the correct age.

If parents accept screening and the blood spot sample is taken, the screening laboratory will perform all processes until screening has been completed for all the conditions – this includes processing initial samples received in the laboratory on or after the child's first birthday and requesting and processing repeat samples if required.

If parents decide not to accept the offer of screening, or the baby is over twelve months old, this should be recorded in clinical notes and on the child health system. Parents should be asked to seek medical advice and remind staff that their baby has not been tested if their baby has signs of chronic health problems such as developmental delay, chronic diarrhoea or repeated lower respiratory tract infections.

If the screen has been declined because although there are no verifiable results available, the family feel the screen was completed elsewhere and a further screen is unnecessary. The Health professional offering the screen should ascertain that they understand that the conditions screened for/testing strategy may differ to that they have already undergone. If the offer of a further screen is still declined, then this should be reported to the SNSL and CHRd as screen declined – previous screen with unverifiable result.

The screening test for CF is unreliable after a baby is 8 weeks old. Older babies with CF may have normal results and so this test will not be done in babies over 8 weeks of age. If a child has repeated chest infections or diarrhoea, medical advice should be sought and a test for cystic fibrosis may be arranged. This test is more complicated than the screening test and so it is not carried out on all babies.

NHS Boards should ensure that there is staff trained and responsible for taking the blood spots in infants that are no longer the responsibility of the midwifery unit. For older babies it may be easier to obtain a venous sample however this must be drawn into a plain syringe with no additives and then the blood should be applied directly to the card. If an automated incision device is used, ensure it is age-appropriate.

12.3 Repeat samples

Informed consent must be taken for all repeat samples. Parents should be informed of the reason for the repeat. To enable parents to make an informed choice about screening for their baby.

Unavoidable repeat samples may be required from a few babies due to prematurity, borderline thyroid stimulating hormone (TSH) results, inconclusive CF screening or having received a blood transfusion. These samples should be taken as soon as possible or at the age directed by the screening laboratory.

A one-week interval between samples is recommended for borderline TSH results to detect any meaningful change in TSH levels. Take a four blood spot sample and mark the blood spot card 'CHT borderline'. Ensure that the 'repeat sample' box is ticked on the blood spot card.

Laboratories may also request a repeat sample due to any of the following:

- Too young for reliable screening which may give rise to a false positive result for CHT
- Too soon after transfusion (<72 hours) therefore metabolite concentrations may not have returned to pre-transfusion levels yet
- Insufficient sample therefore a risk of a false negative result
- Inappropriate application of blood therefore a risk of a false negative or false positive result
- Compressed, damaged or contaminated sample therefore a significant risk of a false negative or inaccurate result
- Day 0 and day 5 sample on same blood spot card therefore unable to determine baby's age at time of sample
- Possible faecal contamination risk of inaccurate CF result
- Incomplete or inaccurate data on the blood spot card, e.g. no/inaccurate CHI number, no/inaccurate date of sample or no/inaccurate date of birth therefore unable to confirm identity of the baby
- Expired blood spot card used
- > 14 days in transit, too old for analysis therefore risk of inaccurate result
- Damaged in transit therefore risk of inaccurate result
- Sickie – too premature for testing therefore risk of inaccurate result

When a repeat sample is requested for any of the above reasons, the sample should be taken within 2 working days of the receipt of the request (unless ongoing transfusions).

If the Child Health Record Department notify the maternity service that a baby either has not had a bloodspot taken/it has not reached the laboratory. The service must action this as soon as possible and complete within **2 working days**. If the family cannot be contacted within this time, then this must be escalated to the Health Board Coordinator or designated deputy. Once the family have been contacted and the sample is taken/or the screen declined the card should be sent **by courier** to the laboratory who should be informed to expect it. This is to avoid any further delay to the already overdue sample being analysed and, should the baby be affected by one of the conditions, still allow time for the child to be identified and commence treatment by day 21, which is a key aim of the programme to reduce the chance of avoidable harm to the baby.