NHS

Screening Programmes

Newborn Blood Spot

Guidelines for Newborn Blood Spot Sampling

Draft Version 0.5 / September 2015

About the NHS Newborn Blood Spot Screening Programme

The NHS Newborn Blood Spot (NBS) Screening Programme screens newborn babies for a number of rare but serious conditions: sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT) and six inherited metabolic diseases (IMDs): phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive) (HCU).

Public Health England (PHE) is responsible for the NHS Screening Programmes. PHE is an executive agency of the Department of Health and works to protect and improve the nation's health and wellbeing, and reduce health inequalities.

NHS Newborn Blood Spot Screening Programme
Floor 2, Zone B
Skipton House
80 London Road
London SE1 6LH
Tel: +44 (0)20 3682 0890
Twitter: @PHE_Screening
www.gov.uk/topic/population-screening-programmes/newborn-blood-spot
www.gov.uk/phe

For queries relating to this document, please contact phe.screeninghelpdesk@nhs.net

Published XXXXX 2015
PHE publications gateway number: XXXXXX

Contents

Introduction

Newborn blood spot (NBS) screening identifies babies who may have rare but serious conditions. The UK National Screening Committee (UK NSC) recommends that all babies are offered screening for sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT) and six inherited metabolic diseases (IMDs): phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive) (HCU). Screening for MSUD, IVA, GA1 and HCU was introduced in England and Wales in January 2015.

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

SCD	severe pain, life-threatening infections and anaemia (symptoms can be present even with treatment)
CF	poor weight gain, frequent chest infections and reduced life expectancy (symptoms can be present even with treatment)
CHT	permanent, serious physical problems and learning disabilities
PKU	permanent, serious learning disabilities
MCADD	serious illness and possible sudden death
MSUD	coma and permanent brain damage or death in some cases
IVA	coma and permanent brain damage or death in some cases
GA1	coma and neurological damage
HCU	learning difficulties, eye problems, osteoporosis, blood clots or strokes

Further information on the conditions is available in the glossary. For further information on all aspects of the newborn blood spot screening programme please visit www.gov.uk/topic/population-screening-programmes/newborn-blood-spot.

These guidelines

These guidelines are written for the screening programme in England. Healthcare professionals in Scotland, Wales and Northern Ireland must be aware of variation in practice and conditions screened for when referring to these guidelines. Units are encouraged to develop local processes in line with these guidelines that demonstrate lines of responsibility.

The guidelines aim to:

- Provide a consistent and clear approach to newborn blood spot sampling
- Support recommendation of newborn blood spot screening to parents
- Support parents in making an informed choice about newborn blood spot screening for their baby
- Support sample takers in obtaining good quality samples to prevent the need for avoidable repeats
- Reduce pain and discomfort during the heel puncture

Additional resources are listed at the end of this document.

Why blood spot quality matters

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 request an ‘avoidable 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 up to eight weeks of age). They are also a waste of healthcare resources (each avoidable repeat costs the NHS around £100). In some cases, parents may refuse to consent to a repeat – this means that the baby will have incomplete screening.

Newborn screening laboratories in England have introduced a national, 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. 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.

In order to take the newborn blood spot sample you will need

- The NHS Screening Programmes' booklet '*Screening tests for you and your baby*'
- Baby's NHS number (use of a bar-coded label is recommended)
- Blood spot card and glassine envelope
- Personal child health record (PCHR) and maternity/professional record
- Water for cleansing
- Non-sterile protective gloves
- Age-appropriate, automated incision device* (manual lancets **must not** be used)
- Sharps box
- Cotton wool/gauze
- Hypoallergenic spot plaster (if required)
- Prepaid/stamped addressed envelope (first class) to despatch sample to screening laboratory (if not using a courier service)

*There is some evidence that an arch-shaped incision device is more effective in providing a good quality sample – see section 3.4.

1. Preparation for taking the blood spot sample

It is important to offer parents an informed choice about screening for their baby, to gain consent and to prepare them for the blood sampling procedure.

Section	Action	Reasoning
1.1	At or prior to antenatal booking, women are given a copy of the *'Screening tests for you and your baby'* booklet. This includes a section on newborn blood spot screening. Ensure parents still have access to the pre-screening booklet at least 24 hours before taking the sample. If not, ensure a copy of the booklet is available to parents. Information on how to order copies of the booklet is available at www.gov.uk/government/collections/population-screening-programmes-leaflets-and-how-to-order-them. Ensure the booklet is in the appropriate language for the parents. Translated versions are available from www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief. If the required language is not available, alternative arrangements should be made with local interpreter services – do not use old translated copies of the booklet.	To enable parents to make an informed choice about screening for their baby. [1-8] The booklet provides information on the conditions screened for, how the sample will be taken and how parents will receive results. It also advises parents on how to prepare for the blood spot (warmth, comfort and feeding of baby).
1.2	Parents should be asked if they have a family history of any of the inherited metabolic diseases.	To ensure a plan is put in place for early testing if appropriate. [9]
1.3	Explain the procedure to parents and record in the maternity/professional record that newborn blood spot screening has been discussed and recommended, the booklet given and consent sought.	Good record keeping is an integral part of nursing and midwifery practice. [10-12]

	Verbal consent is adequate.	
1.4	Parents should be asked if they wish to be contacted about research linked to the screening programme. Information is available at www.nhs.uk/Conditions/pregnancy-and-baby/Pages/newborn-blood-spot-cards.aspx. If a parent does not wish to be contacted about future research on their baby's newborn blood spot screening sample, **'No research contact'** should be recorded clearly on the blood spot card. Ensure parents are aware that patient identifiable information may be stored by the NHS Sickle Cell and Thalassaemia Screening Programme. Information is available at www.gov.uk/newborn-outcomes-project-definition-and-implementation.	Stored blood spot cards are used to monitor and improve the newborn screening programme. In accordance with the Code of Practice for the Retention and Storage of Residual Spots. [13] This is used to monitor and improve screening for sickle cell and thalassaemia. The use of patient identifiable information obtained from sickle cell and thalassaemia screening was approved by the National Information Governance Board. This is reviewed annually by the NHS Health Research Authority Confidentiality Advisory Group. [14]
1.5	**If the parents consent to screening:** Record the parents' decision as 'consent' in the PCHR and the maternity/professional record and proceed with taking the sample. If the baby is in hospital, the parents' consent decision should also be recorded in the baby's hospital records.	By recording information in the PCHR, parents and other health professionals will have information about the status of the baby in relation to the screening test.
1.6	The blood spot sample should be taken on day 5* for all babies regardless of medical condition, medication, milk feeding and prematurity. **For the purpose of screening, day of birth is day 0** (some information systems record day of birth as day 1).	To enable timely detection of abnormal results and initiation of appropriate treatment.

	Arrange a convenient time to take the blood spot sample on this day. *In exceptional circumstances the sample can be taken between day 5 and day 8.	To ensure parents are aware of when the newborn blood spot screening test will happen. For example, if the baby has had a blood transfusion (see section 5.5).
1.7	Parents may decline screening for SCD, CF and CHT individually but the six IMDs can only be declined as a group.	The screening laboratory tests for all the IMDs using one punched disc (see section 3.6).
1.8	**If the parents decline screening:** For each condition declined, record the decline and reason (if stated) in the PCHR ('birth details' section) and maternity/professional record. Complete the blood spot card as described in section 2 and send the completed card marked as **'Decline'** to the screening laboratory. The sample taker is to inform the GP and health visitor (if applicable) of the conditions declined – template letters are available at www.gov.uk/government/publications/declined-newborn-blood-spot-screening-template-letters. A template letter is also available for informing the CHRD directly. It is best practice to also inform the NBS lead midwife/manager.	To monitor rates of consents / declines and effectively communicate parents' requests to the laboratory and child health records department (CHRD). To also prevent the re-offer of screening. To ensure the family's GP does not assume testing has been completed and thereby, should symptoms arise, rule out the possibility of an affected child.
1.9	A template letter is also available to complete and give to parents if they decline screening – see Appendix 1. Inform parents whom to contact if they change their mind or would like further information. Record this information in the PCHR.	To provide parents with written confirmation of their decision and information on the possible consequences of their baby not being screened. To ensure parents know how to have their baby screened if they wish.

2. Entering the details on the blood spot card

The baby's NHS number on the blood spot card is mandatory in England. Use of a bar-coded label is recommended. This saves health professionals' time in data entry and minimises transcription errors. NHS number bar-coded labels should be generated at the point of notification of birth and given to parents with the PCHR on transfer from hospital to home or before, so that they are available for blood spot screening.

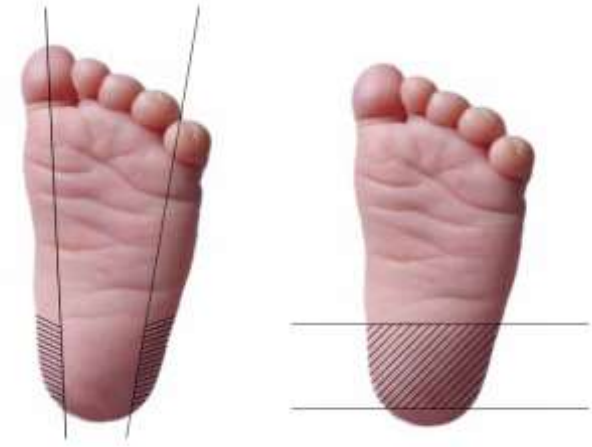
Section	Action	Reasoning
2.1	Check expiry date on the front of the blood spot card.	The laboratory will be unable to process the sample if the blood spot card is out of date, and a repeat sample will be required, resulting in a possible delay in treatment.
2.2	Complete the details on the blood spot card **at the time of sampling**. Use of a bar-coded label is recommended. **When using a bar-coded label:** • Ensure that no sections of the bar-code or text are cut off or missing • Check with the parents that all details on the label are correct and make any necessary changes • Do not use incomplete / unreadable labels. Instead, complete the details on the blood spot card in legible handwriting – see **'If label is not available'** (below) • Apply one label to each sheet of the blood spot card at the time of sampling (**do not** apply in advance of the test) **Using legible handwriting, complete all fields on the blood spot card that are not included on the bar-coded label.**	To ensure the label meets NBS screening programme criteria. [15] If the laboratory is unable to read the information on the blood spot card or the card is not fully/accurately completed, the sample will not be processed and the baby will require a repeat sample and may have treatment delayed. This may cause anxiety and distress to families.

	If label is not available: Ensure ALL fields are completed using legible handwriting. Record the maternity organisation code in the PCT field. Further information on bar-coded labels can be found at www.gov.uk/government/publications/barcode-labels-quality-assurance-in-newborn-blood-spot-screening.	All information on the blood spot card is required by the laboratory. This will help the laboratories to collect accurate avoidable repeat data.
2.3	When completing the blood spot card care must be taken to place the card on a clean surface.	To avoid contaminating the blood spot sample.
2.4	Record any of the following in the 'comments' box on the blood spot card: • Baby's known medical condition • Family history relevant to the conditions screened for • Reason for sample if not taken on day 5-8 (e.g. pre-transfusion, preterm CHT)	To assist the newborn screening laboratory with linking antenatal and newborn screening results. To ensure the result is interpreted correctly.
2.5	Check the completed blood spot card with the parents and make any necessary changes.	To ensure that the baby's and mother's details are accurate before collecting the blood spot sample.

3. Collecting the blood spot sample

Section	Action	Reasoning
3.1	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. [16-17] Engaging the baby through face-to-face contact, voice and touch may be beneficial. Suggest the baby is breast feeding during the heel prick as an analgesic. [18-21] An alternative to breast feeding is to offer expressed breast milk, non-nutritive sucking (e.g. a pacifier) or a sucrose or glucose solution (if available). [19-23] Whilst there is no evidence that formula feed has analgesic properties, parents may comfort formula-fed babies with a feed during the procedure.	 To make it easier for the baby to regain his or her calm and cope with the procedure. To reduce the pain/discomfort of the procedure. Painful procedures are a medical indication for use of pacifiers or sweet solutions. This does not undermine UNICEF UK's *Baby Friendly Initiative standards.* [24]
3.2	Clean the heel by washing thoroughly with plain water using cotton wool/gauze. 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.	Contamination of the sample may affect the test results. The NHS Newborn Blood Spot Screening Programme has received reports of babies being scalded/burned during warming of the heel in preparation for blood spot sampling. [25, 26] Soap or detergent can irritate infantile skin. [27] Faeces contain very high concentrations of immunoreactive trypsinogen (IRT) (IRT is

		measured during screening for cystic fibrosis). Faecal contamination may lead to a false-positive result.
	Do not use alcohol or alcohol wipes.	The use of alcohol for skin preparation in neonates and premature infants can cause burns and blisters. [27-31]
	The heel should be completely dry before taking the sample. Soft paraffin solutions such as Vaseline® should not be used for heel punctures.	To comply with infection control guidelines. [32] Paraffin solutions can alter the results of the blood spot test and can clog the equipment used.
3.3	Wash hands and apply gloves.	Universal precaution before taking a blood sample. [32-33]
3.4	Ensure the baby is warm and comfortable. **Warming of the foot is not required.**	There is no evidence that warming aids blood flow. [34, 33, 38]
	Obtain the sample using an age-appropriate automated incision device (different lancets are available for different ages). [34-36] There is some evidence that an arch-shaped incision device is more effective in providing a good quality sample, reducing the number of heel punctures per sample, the time taken to complete the sample, bruising, the time the baby cried, and the need to repeat the sample. [34-35] Manual lancets **must not** be used.	Automated incision devices reduce pain and bruising, allow users to obtain the sample more quickly and reduce the risk of accidental injury from manual lancets. [34, 39]
	For full-term and preterm infants, the external and internal limits of the calcaneus are the preferred puncture site. This is marked by the shaded areas in diagram A. Skin puncture must be no deeper than 2.0 mm. For infants who have had repeated heel	The skin to calcaneus depth is greater in these areas. To minimise the risk of

	punctures, the areas marked in diagram B may also be used. When using the whole plantar surface, an automated incision device with a penetrative depth of no more than 1.0 mm is recommended. [37] Avoid posterior curvature of the heel. Allow the heel to hang down to assist blood flow. Before activation place the automated incision device against the heel in accordance with manufacturers' instruction.	calcaneal puncture that may lead to calcaneal osteomyelitis (inflammation of the heel bone). [37, 40] This reduces the soft tissue damage and pain from repeated heel puncture in the same area. 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.
3.5	Adapted from Jain & Rutter [41] Diagram A For full-term and preterm infants Diagram B For infants who have had repeated heel punctures 	
3.6	**Good quality blood spots are vital for ensuring that babies with rare but serious conditions are identified and treated early.**	Evidence shows that poor quality samples could lead to a false-negative or false-positive result which means that a baby with a serious condition might be missed

		or referred for further tests unnecessarily.
	The aim is to fill each circle on the newborn blood spot card, using a **single** drop of blood (see diagram C).	From each circle the laboratory punches out several small discs.
	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. The first drop of blood should be used.	The sample needs to be sufficient to screen for all of the conditions and to be used for further testing if required e.g. to check a screen-positive results.
	There is no need to collect additional blood for expanded screening.	No extra punches are needed to perform the new tests.
	Do not squeeze the foot in an attempt to increase blood flow.	This can cause pain and bruising to the baby. [17, 32, 33]
	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 four circles completely. Always ensure that the sample is taken from the front of the card and not from the back. Spots that exceed the dotted lines on the filter paper are acceptable provided that a single drop of blood has been used.	This gives the optimum amount of blood for the laboratory to utilise.
	Do not compress or apply pressure to the blood spots (for example when sealing the postage envelope).	Applying pressure reduces the density of blood on the sample – there is significant risk that this could lead to a ‘suspected’ result being missed (see diagram C).

3.7 Diagram C

Correct	Reasoning
A single, evenly saturated drop of blood that fills the circle completely and soaks through to the back of the blood spot card 	Good quality blood spots are essential to obtain accurate screening results – **this prevents babies with a condition being missed or being referred for further tests unnecessarily**
Incorrect	**Reasoning**
Insufficient sample (e.g. one small spot of blood) 	Risk of false-negative result
Multispotted sample (e.g. several small spots of blood) 	Risk of false-negative result
Layered sample (e.g. one spot of blood is layered directly on top of another or blood is applied on both sides of the blood spot card) 	Risk of false-positive result

	Blood not soaked through to back of blood spot card Front of card  Back of card 	Risk of false-negative result
	Compressed sample  (can be identified through staining of the envelope) 	Significant risk of false-negative result
	Contaminated sample 	Might affect the test results
3.6	**If the blood flow ceases:** The congealed blood should be wiped away firmly with cotton wool or gauze. Gently ‘massage’ the foot, avoid squeezing, and drop the blood onto the blood spot card.	To disturb the clot and encourage blood flow. To reduce the amount of pain and bruising caused by the procedure.

3.7	**If the baby is not bleeding, a second puncture is necessary:** The second puncture should be performed on a different part of the same foot or on the other foot, as marked by the shaded areas in diagrams A and B.	The original site is avoided to prevent the sample from containing excessive tissue fluid and to reduce pain.
3.8	When the sample collection is complete, wipe excess blood from the heel and apply gentle pressure to the wound with cotton wool or gauze.	To prevent excessive bleeding and bruising and to protect the wound.
3.9	Apply a hypoallergenic spot plaster if required and remind the parent to remove the plaster in a few hours.	A small minority of babies may react adversely to a hypoallergenic plaster when left on for too long, which can cause distress.

4. After taking the blood spot sample

It is important that the laboratory receives the blood sample promptly to ensure that screen-positive babies are seen quickly. Parents also need to know when to expect the results. This will help to reduce their concerns about the results, as well as provide an additional safety net in following up missing results.

Section	Action	Reasoning
4.1	Check the completed blood spot card with the parents and make any necessary changes.	To ensure that the baby's and mother's details are accurate before the sample is despatched.
	Allow blood spots to air-dry away from direct sunlight or heat before placing in the glassine envelope.	Wet samples can stick to the envelope and a repeat sample will be required.
	Despatch the blood spot card in the prepaid/stamped addressed envelope on the same day (if not using courier). If not possible, despatch within 24 hours of taking the sample. Despatch should not be delayed in order to batch blood spot cards together for postage. If a post box is used, ensure that it is one that is emptied frequently.	Ensures that the blood spot card is received in laboratory within three working days of the sample being taken. Timeliness of despatch enables early analysis and subsequent treatment.
	Central validation of samples (for example in the maternity unit) is not recommended.	Central validation can cause delayed despatch and generate false avoidable repeat data.
	Provider organisations, in agreement with their regional newborn screening laboratory, should have contingency plans in place for any possible exceptional circumstances that may delay samples reaching the laboratory in time, i.e. postal strikes, severe weather disruptions.	Laboratories will reject samples if received 14 or more days after the sample was taken.
	Record date, method, blood spot card serial number and location of sample despatch, as per local protocol. If a post box is used, record its post code (visible on each box).	For internal audit purposes, and to provide a cross-check between sample taker and laboratory.
4.2	Record that the sample has been taken in the PCHR and the maternity/professional record, complying with local protocols.	To comply with NMC record keeping guidelines. [9-11]

	Record and notify screening status on discharge / transfer notifications.	To ensure that screening status is known and to transfer responsibility for obtaining any outstanding tests (in accordance with local pathway).
4.3	Inform parents that they will receive the results within 6-8 weeks of their baby's birth [42]. If the baby screens positive for a condition the parents will be contacted sooner (please see *'Screening tests for you and your baby'* for further details). Inform parents how they will receive the results, e.g. by post or via the health visitor, as per local policy. Ensure that parents know to contact their health visitor if results are not received within 6-8 weeks.	To ensure all parents receive results of screening. Note that the programme does not have access to screening results.

5. Special circumstances: Babies born preterm or cared for in hospital specialist units

Some babies will be in hospital when their blood spot sample is due to be taken. This section highlights the needs of babies who are cared for in neonatal units (this includes Paediatric Intensive Care Units, Neonatal Intensive Care Units, Special Care Baby Units, cardiac units, surgical units, transition wards, etc.), preterm babies born at less than 32 weeks (less than or equal to 31 weeks + 6 days) and those who experience multiple blood spot samples taken from the heel.

Section	Action	Reasoning
5.1	Babies admitted to neonatal units are likely to have multiple blood samples taken. Blood spot screening should be coordinated with other tests when possible. 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.	 To minimise the number of invasive procedures. Contamination with EDTA can affect newborn screening results. Lithium heparin is difficult to detect and can affect DNA testing. This could affect the protocol used to detect CF and SCD.
5.2	Babies less than 5 days of age should have a **single** circle blood spot sample taken on admission/prior to blood transfusion for the routine screening test for SCD. This should be on a separate blood spot card marked **'Pre-transfusion'.** Complete the details on the blood spot card as described in section 2. Tape or a sticky label can be placed over the three unused circles.	The screening test for SCD cannot be performed on the routine day 5 sample if the baby has received a blood transfusion before the sample has been taken. To avoid the day 5 sample being added to the pre-transfusion blood spot card.

5.3	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 day 5 sample if the baby has received a blood transfusion in the interim. 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 day 5 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.	To prevent the need for DNA analysis to complete SCD screening (see section 5.6). To ensure new unit is aware that the pre-transfusion sample has been taken.
5.4	The routine blood spot sample (four spots) should be taken on day 5* for all babies regardless of medical condition, medication, milk feeding and prematurity. **For the purpose of screening, day of birth is day 0** (some information systems record day of birth as day 1). *In exceptional circumstances the sample can be taken between day 5 and day 8. Complete the details on blood spot card as described in section 2.	To enable timely detection of abnormal results and initiation of appropriate treatment.
5.5	When a baby has had a blood transfusion, 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 and the IMDs. (For intrauterine transfusion count day of birth as date of transfusion). However, in the event of multiple blood transfusions, 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 enable metabolite concentrations to return to pre-transfusion levels. To ensure all babies are screened by day 8 regardless of blood transfusion status.

	If there has not been an interval of at least 3 days (72 hours) between the last transfusion and the routine sample, a repeat sample should be taken at least 3 days (72 hours) after the last transfusion. The date of the last blood transfusion before the blood spot must be recorded on the blood spot card and on discharge / transfer notifications. Please refer to sections 5.2. 5.3 and 5.6 for SCD. See Appendix 2 for a flowchart and scenarios.	To reduce the chance of missing a baby with one of the conditions. To permit appropriate interpretation of results. To aid interpretation of the guidelines.
5.6	If a baby has not had a pre-transfusion sample taken, the laboratory may forward the routine day 5 sample to the DNA laboratory for analysis as a failsafe. An additional cost for this may be incurred by the Trust. Further information is available at www.gov.uk/government/publications/dna-tests-for-transfused-babies-sickle-cell-and-thalassaemia-screening.	To ensure all babies are screened for SCD.
5.7	An assessment of the baby's level of distress and ability to tolerate handling must be made before initiating comfort measures. [43] Where appropriate for the baby's condition, analgesia and comfort measures may be used as described in section 3.1.	To reduce the pain/discomfort of the procedure.
5.8	Inform parents of any outstanding screening tests, and record this in the PCHR. Advise parents which healthcare professional will be responsible for completing the blood spot screening for their baby and approximately when it will occur. 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	To ensure that all babies are screened.

	neonatal period. The screening status of the baby is to be recorded on an auditable IT system and in the discharge/transfer documentation.	
CHT screening for preterm infants		
5.9	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 day 5 sample (counting day of birth as day 0). 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.	To ensure a valid sample for congenital hypothyroidism screening as immaturity can mask this condition.
	See Appendix 3 for a list of possible scenarios (including when a baby has had a blood transfusion).	To enable interpretation of the policy.
	Complete the details on the blood spot card as described in section 2, recording 'CHT preterm' on the blood spot card. Write the gestational age on the card.	To ensure laboratory is aware of reason for second sample.
	If the baby is being discharged home before 28 days of age, write **'discharged home'** on blood spot card. Two spots on the blood spot card should be filled with blood.	To ensure laboratory knows why 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.
	In babies who are transferred before they reach 28 days of age, the responsibility for completing screening is transferred to healthcare professionals in the receiving unit.	To ensure screening will be completed by receiving unit.
	Record all blood spot samples taken in baby's hospital records, on transfer documentation, PCHR and on an auditable IT system.	To ensure all babies born at less than 32 weeks (equal to or less than 31 weeks + 6 days) are screened.

6. Ensuring completeness of coverage of newborn screening

Section	Action	Reasoning
Older babies		
6.1	Babies up to 12 months of age who become the responsibility of the provider organisation should be offered screening for all nine conditions if there are no documented results (or declines) for SCD, CF, CHT, PKU and MCADD. If there are documented results for these five conditions no further offer of screening is required. Babies can only be screened for CF up to 56 days of age. Sample taker to inform GP if baby not screened for CF for this reason. Only written confirmation of conclusive results (that you can interpret) should be accepted. All reasonable attempts should be made to find the results; however, this should not unduly delay screening. Provider organisations should ensure that they have easy access to staff trained and responsible for taking blood spots in infants when they are no longer the responsibility of the midwifery unit. If the conclusive results cannot be found, a sample should be taken, using the blood spot card (completed as described in section 2) and sent to the screening laboratory. Either a	To identify any affected baby and ensure treatment commences as soon as possible [44, 45]. The routine screening test for CF (IRT) is no longer reliable after 56 days of age. To ensure the family's GP does not assume testing for CF has been completed and thereby, should symptoms arise, rule out the possibility of an affected child. The screening programme does not maintain a record of the conditions screened for in other countries. Venepuncture, when taken by a skilled phlebotomist, is less painful than heel prick; however this may be technically difficult in babies. [46, 47]

	capillary or venous sample can be spotted onto the blood spot card. If an automated incision device is used, ensure it is age-appropriate (different lancets are available for different ages). Record clearly on the blood spot card the method of sample taking. Local policy is to stipulate how many attempts to contact the family should be made over a specified timeframe before recording 'not screened'. Sample taker to inform GP and CHRD. **If the parents decline screening:** For each condition declined, record the decline and reason (if stated) in the PCHR ('birth details' section) and maternity/professional record. Complete the blood spot card as described in section 2 and send the completed card marked as **'Decline'** to the screening laboratory. The sample taker is to inform the GP and health visitor (if applicable) of the conditions declined – template letters are available at www.gov.uk/government/publications/declined-newborn-blood-spot-screening-template-letters. A template letter is also available for informing the CHRD directly. A template letter is also available to complete and give to parents if they decline screening – see Appendix 1.	To monitor rates of consents / declines and effectively communicate parents' requests to the laboratory and CHRD. To also prevent the re-offer of screening. To ensure the family's GP does not assume testing has been completed and thereby, should symptoms arise, rule out the possibility of an affected child. To provide parents with written confirmation of their decision and information on the possible consequences of their baby not being screened.
Repeat samples		
6.2	Informed consent must be taken for all repeat samples (see section 1). Parents should be informed of the reason for the repeat.	To enable parents to make an informed choice about screening for their baby [1-8].

	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.	To ensure screened babies receive a valid result.
	A one week interval between samples is recommended for borderline TSH results. Take a four blood spot sample and mark the blood spot card 'CHT borderline'.	An interval of one week is required to detect any meaningful change in TSH levels.
	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 [48]:	
	- Too young for reliable screening	May give rise to a false-positive result for CHT.
	- Too soon after transfusion (<72 hours)	Metabolite concentrations may not have returned to pre-transfusion levels.
	- Insufficient sample	Risk of false-negative result.
	- Inappropriate application of blood	Risk of false-negative or false-positive result.
	- Compressed, damaged or contaminated sample	Significant risk of false-negative result / risk of inaccurate result.
	- Day 0 and day 5 sample on same blood spot card	Unable to confirm baby's age at sample.
	- Possible faecal contamination	Risk of inaccurate CF result.
	- Incomplete or inaccurate data on the blood spot card, e.g. no/inaccurate NHS number, no/inaccurate date of sample or no/inaccurate date of birth	Unable to confirm identity of baby.
	- Expired blood spot card used	Risk of inaccurate result.
	- > 14 days in transit, too old for analysis	Risk of inaccurate result.

	- Damaged in transit - Sickle – too premature for testing When a repeat sample is requested for any of the above reasons, the sample should be taken within 72 hours of the receipt of the request (unless ongoing transfusions).	Risk of inaccurate result. Risk of inaccurate result.
Failsafe processes		
6.3	Provider organisations should ensure failsafe arrangements are in place for notifying screening status when the care of a baby is transferred. This includes babies who are transferred in the neonatal period or discharged home before screening for all tests is complete.	To ensure all babies eligible for screening are screened, all positive babies receive timely treatment and parents receive their results by 6-8 weeks of age. [8]
	Provider organisations should implement failsafe measures to ensure that: • All eligible babies are identified • All identified babies are offered screening • All babies, whose parents accept the offer of screening, are screened • All samples are received in the screening laboratory • All positive babies receive treatment within national standards • Parents receive the results by 6-8 weeks of age	To prevent irreversible harm that can be caused to babies affected by the screened conditions when samples are delayed or are not received by laboratories.
	The screening status of all eligible babies should be recorded on an auditable child health IT system. Provider organisations should also perform daily checks of the Newborn Blood Spot Failsafe Solution (NBSFS) to identify babies that might have missed newborn blood spot screening. For	To ensure all eligible babies are offered screening and are screened.

	more information on the NBSFS see www.gov.uk/government/publications/newborn-blood-spot-screening-failsafe-procedures.	

Monitoring blood spot quality

Blood spot quality is monitored regularly through the collection of data against key performance indicator (KPI) NB2 (quarterly) [49] and blood spot standard 6 (annually) [42]:

- Acceptable level: the avoidable blood spot repeat rate is less than or equal to 2%
- Achievable level: the avoidable blood spot repeat rate is less than or equal to 0.5%

Data is collated and published by the NHS Screening Programmes' KPI team and the NBS programme. The Screening Quality Assurance Service monitors the data to check that the standard is being met and encourages continuous improvement.

Additional resources

- Online learning modules on improving blood spot quality and expanded newborn screening: cpd.screening.nhs.uk/bloodspot-elearning
- Short films, including a video of a community midwife discussing how she improved her avoidable repeat rate: cpd.screening.nhs.uk/newbornbloodspot
- Interactive blood spot card: cpd.screening.nhs.uk/interactivecard.php
- Standards for newborn blood spot screening [42]

There may also be **local initiatives** to support you in taking good quality blood spot samples – speak to your local screening coordinator or regional quality assurance team.

References

1. National Institute for Clinical Excellence (2008) *Antenatal Care: NICE guidelines [CG62]* [Online] Available at: http://www.nice.org.uk/guidance/cg62 (accessed 08 July 2015).

2. Department of Health (2009) *Reference Guide to Consent for Examination or Treatment, 2nd edition* [Online] Available at: https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/138296/dh_103653__1_.pdf (accessed 08 July 2015).

3. Department of Health/Partnerships for Children, Families and Maternity (2007) *Maternity matters: choice, access and continuity of care in a safe service* [Online] Available at: http://webarchive.nationalarchives.gov.uk/20130107105354/http:/www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_074199.pdf (accessed 08 July 2015).

4. Detmar, S., et al. (2007) *Information and informed consent for neonatal screening: opinions and preferences of parents.* Birth, 34(3): p. 238-44.

5. Parsons, E.P., et al. (2007) *Mothers' accounts of screening newborn babies in Wales (UK).* Midwifery, 23(1): p. 59-65.

6. Davis, T.C., et al. (2006) *Recommendations for effective newborn screening communication: results of focus groups with parents, providers, and experts.* Pediatrics, 117(5 Pt 2): p. S326-40.

7. Pittam, G. and Allaby, M. (2012) *A report for the UK National Screening Committee Informed Consent in Antenatal and Newborn Screening*. Solutions for Public Health: Oxford.

8. NHS England (2014) *NHS public health Functions agreement 2015-16 Service specification no.19, NHS Newborn Blood Spot Screening Programme* [Online] Available at: https://www.gov.uk/government/publications/public-health-commissioning-in-the-nhs-2015-to-2016 (accessed: 08 July 2015).

9. NHS Newborn Blood Spot Screening Programme (2015) *A laboratory guide to newborn blood spot screening for inherited metabolic diseases* [Online] Available at: https://www.gov.uk/government/publications/newborn-blood-spot-screening-laboratory-guide-for-imds (accessed 06 August 2015).

10. Nursing and Midwifery Council (2012) *Midwives Rules and Standards* [Online] Available at: http://www.nmc.org.uk/standards/additional-standards/midwives-rules-and-standards/ (accessed 08 July 2015).

11. Nursing and Midwifery Council (2009) *Record Keeping: Guidance for Nurses and Midwives,* [Online] Available at: http://www.nipec.hscni.net/Image/SitePDFS/nmcGuidanceRecordKeepingGuidanceforNursesandMidwives.pdf (accessed 08 July 2015).

12. Nursing and Midwifery Council (2015) *The Code: Professional standards of practice and behaviour for nurses and midwives* [Online] Available at: http://www.nmc.org.uk/globalassets/sitedocuments/nmc-publications/revised-new-nmc-code.pdf (accessed 06 August 2015).

13. UK Newborn Screening Programme Centre (2005) *Code of Practice for the Retention and Storage of Residual Spots* [Online] Available at: https://www.gov.uk/government/publications/newborn-blood-spot-screening-code-of-practice-for-the-retention-and-storage-of-residual-spots (accessed 08 July 2015).

14. Department of Health (2006) *The National Health Service Act 2006, The National Health Service (Wales) Act 2006 and The National Health Service (Consequential Provisions) Act 2006*: London.

15. NHS Newborn Blood Spot Screening Programme / NHS Connecting for Health (2014) *NHS Numbers for Newborn Screening: Output Based Specification for the Blood Spot Card Label v3.4* [Online] Available at: https://www.gov.uk/government/publications/nhs-numbers-for-newborn-screening-specification-for-the-blood-spot-card-label (accessed 06 August 2015).

16. Morrow, C., Hidinger, A. and Wilkinson-Faulk, D. (2010) *Reducing neonatal pain during routine heel lance procedures.* MCN Am J Matern Child Nurs, 35(6): p. 346-54; quiz 354-6.

17. Shepherd, A., Glenesk, A. and Niven, C. (2006) *What influence does experience play in heel prick blood sampling?* J Neonatal Nursing, 12(3).

18. Shah, P.S., Aliwalas, L.I. and Shah, V. (2006) *Breastfeeding or breast milk for procedural pain in neonates.* Cochrane Database Syst Rev, 3: p. CD004950.

19. Yilmaz, F. and Arikan, D. (2011) *The effects of various interventions to newborns on pain and duration of crying.* J Clin Nurs, 20(7-8): p. 1008-17.

20. Codipietro, L., Ceccarelli, M. and Ponzone, A. (2008) *Breastfeeding or oral sucrose solution in term neonates receiving heel lance: a randomized, controlled trial.* Pediatrics, 122(3):p. e716-21.

21. Uga, E., et al. (2008) *Heel lance in newborn during breastfeeding: an evaluation of analgesic effect of this procedure.* Ital J Pediatr, 34(1): p. 3.

22. Taddio, A., Shah, V. and Katz, J. (2009) *Reduced infant response to a routine care procedure after sucrose analgesia.* Pediatrics, 123(3): p. e425-9.

23. Stevens, B., Yamada, J. and Ohlsson, A. (2010) *Sucrose for analgesia in newborn infants undergoing painful procedures.* Cochrane Database Syst Rev, p. CD001069.

24. UNICEF UK (2012) *Baby Friendly Initiative standards* [Online] Available at: http://www.unicef.org.uk/Documents/Baby_Friendly/Guidance/Baby_Friendly_guidance_2012.pdf (accessed 09 July 2015).

25. Ray, R., Godwin, Y. and Shepherd, A. (2011) *Convective burn from use of hairdryer for heel warming prior to the heel prick test - a case report.* BMC Pediatr, 11: p. 30.

26. Hassan, Z. and Shah, M. (2005) *Scald injury from the Guthrie test: should the heel be warmed?* Arch Dis Child Fetal Neonatal Ed, 90(6): p. F533-4.

27. Paediatric Formulary Committee (2015) *BNF for Children* [Online] London: BMJ Group, Pharmaceutical Press, and RCPCH Publications Available at: http://www.evidence.nhs.uk/formulary/bnfc/current/ (accessed 24 August 2015).

28. O'Grady, N.P., et al. (2011) *Summary of recommendations: Guidelines for the Prevention of Intravascular Catheter-related Infections.* Clin Infect Dis, 52(9): p. 1087-99.

29. Scales, K. (2009) *Correct use of chlorhexidine in intravenous practice.* Nursing Standard, 24(8): p. 41-47.

30. Committee on Infectious Diseases (2007) *Infection prevention and control in pediatric ambulatory settings.* Pediatrics, 120(3): p. 650-65.

31. Reynolds, P.R., Banerjee, S. and Meek, J.H. (2005) *Alcohol burns in extremely low birthweight infants: still occurring.* Arch Dis Child Fetal Neonatal Ed, 90(1): p. F10.

32. Pratt, R.J., et al. (2007) *epic2: National evidence-based guidelines for preventing healthcare associated infections in NHS hospitals in England.* J Hosp Infect, 65 Suppl 1: p. S1-64.

33. National Institute for Health and Care Excellence (2012), *Infection: Prevention and control of healthcare-associated infections in primary and community care NICE clinical guideline 139* [Online] Available at: https://www.nice.org.uk/guidance/cg139/resources/guidance-infection-pdf (accessed 09 July 2015).

34. Glenesk, A., Shepherd, A. and Niven, C. *Blood spot testing: comparing techniques and automated devices.* Br J Midwifery, 14(1).

35. Shepherd, A.J., et al. (2006) *A Scottish study of heel-prick blood sampling in newborn babies.* Midwifery, 22(2): p. 158-68.

36. Vertanen, H., et al. (2001) *An automatic incision device for obtaining blood samples from the heels of preterm infants causes less damage than a conventional manual lancet.* Arch Dis Child Fetal Neonatal Ed, 84(1): p. F53-5.

37. Arena, J., et al. (2005) *Skin to calcaneus distance in the neonate.* Arch Dis Child Fetal Neonatal Ed, 90(4): p. F328-f331.

38. Naughten, F. (2005) *The heel prick: how efficient is common practice?* RCM Midwives, 8(3): p.112-4.

39. Janes, M., et al. (2002) *Comparison of capillary blood sampling using an automated incision device with and without warming the heel.* J Perinatol, 22(2): p. 154-8.

40. Yuksel, S., et al. (2007) *Osteomyelitis of the calcaneus in the newborn: an ongoing complication of Guthrie test.* Eur J Pediatr,166(5): p. 503-4.

41. Jain, A. and Rutter, N. (1999) *Ultrasound study of heel to calcaneum depth in neonates.* Arch Dis Child Fetal Neonatal Ed, 80(3): p. F243-5.

42. NHS Newborn Blood Spot Screening Programme (2013) *Standards for Newborn Blood Spot Screening* [Online] Available at: https://www.gov.uk/government/publications/standards-for-nhs-newborn-blood-spot-screening (accessed 07 July 2015).

43. Castral, T.C., et al. (2008) *The effects of skin-to-skin contact during acute pain in preterm newborns.* Eur J Pain, 12(4): p. 464-71.

44. NHS England (2014) *2015-16 National Heath Visiting Core Service Specification* [Online] Available at: http://www.england.nhs.uk/wp-content/uploads/2014/12/hv-serv-spec-dec14-fin.pdf (accessed 06 August 2015).

45. NHS Newborn Blood Spot Screening Programme (2015) *Procedures for newborn blood spot (NBS) screening of babies under a year of age, for whom screening results are not available* [Online] Available at: https://www.gov.uk/government/publications/movers-in-screening-babies-with-no-available-records (accessed 06 August 2015).

46. Shah, V. and Ohlsson, A. (2007) *Venepuncture versus heel lance for blood sampling in term neonates.* Cochrane Database Syst Rev, (4) p. CD001452.

47. Ogawa, S., et al. (2005) *Venepuncture is preferable to heel lance for blood sampling in term neonates.* Arch Dis Child Fetal Neonatal Ed, 90(5): p. F432-6.

48. NHS Newborn Blood Spot Screening Programme (2014) *Status codes: version 4.2* [Online] Available: https://www.gov.uk/government/publications/status-codes-for-the-newborn-blood-spot-nbs-screening-programme (accessed 06 August 2015).

49. NHS Screening Programmes (2015) *NHS screening programmes: national data reporting* Available at: https://www.gov.uk/government/collections/nhs-screening-programmes-national-data-reporting (accessed 06 August 2015).

Appendices

Appendix 1: Template letter for parents that decline newborn blood spot screening

Dear Parent/Carer

Re: Declined newborn blood spot screening – [baby's name, date of birth, NHS number]

Born and resident babies – delete as applicable: *I am writing to confirm that you would like to decline the offer of newborn blood spot screening for your baby.*

Movers in – delete as applicable: *I am writing to confirm that we do not have a record of your baby's newborn blood spot screening results and that you would like to decline the offer of screening in our area. Newborn blood spot screening is offered to all babies born in the UK and those that move in under 12 months of age without screening results.*

Newborn blood spot screening involves taking a blood sample to find out if your baby has one of several rare but serious health conditions. If these conditions are detected early, they can be treated effectively. However, if they are not detected, they may cause irreversible harm to your child. Screening is not compulsory, but it's strongly recommended because it could save your baby's life.

Information on the conditions screened for is available in the '*Screening tests for you and your baby*' booklet (www.gov.uk/government/publications/screening-tests-for-you-and-your-baby-description-in-brief) and on NHS Choices (www.nhs.uk/Conditions/pregnancy-and-baby/Pages/newborn-blood-spot-test.aspx).

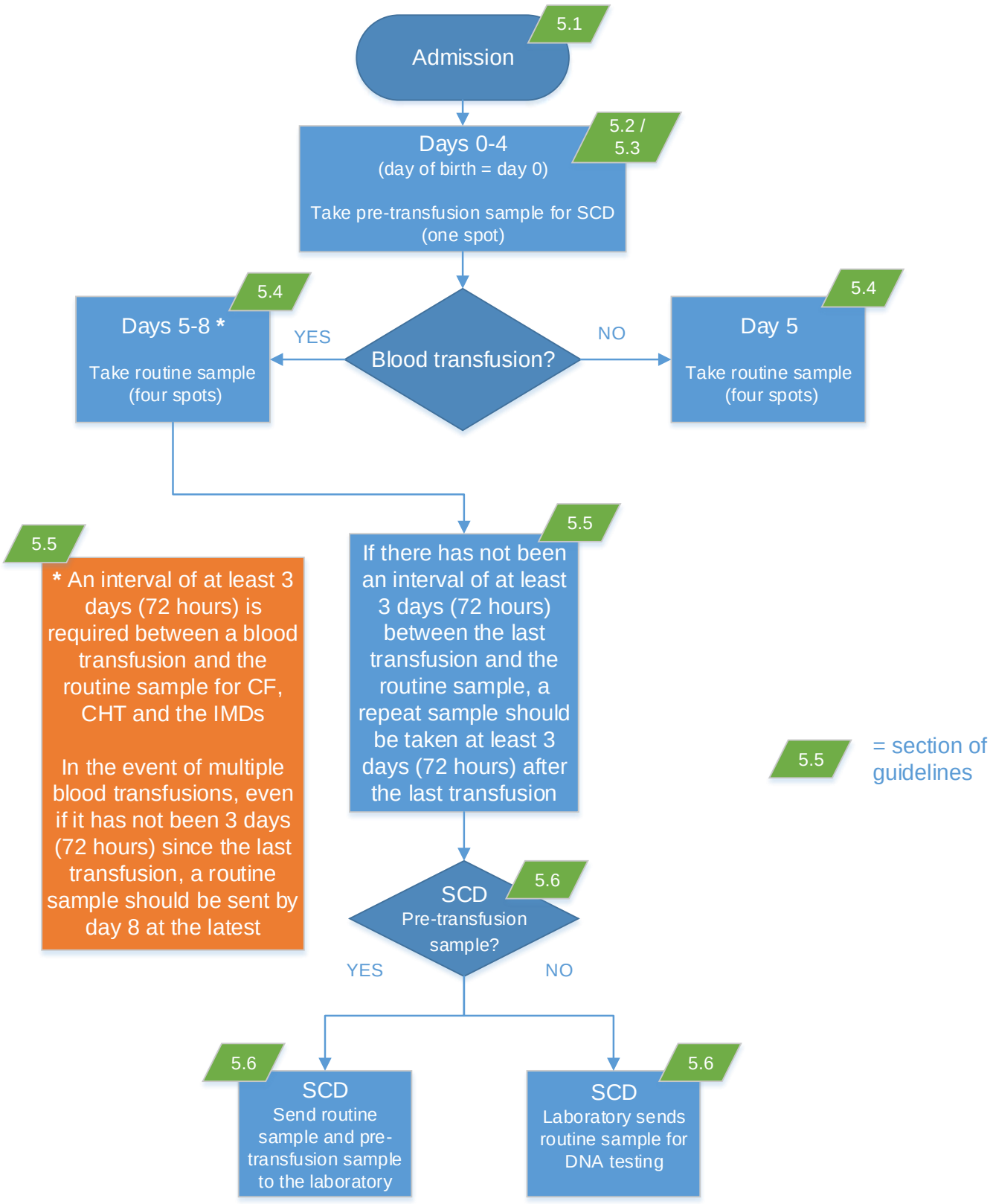
If you change your mind

We understand that you have the right to decline screening for your baby and will record this in your baby's health records. However, if you change your mind, babies can be screened up to the age of 12 months for all the conditions except cystic fibrosis. Cystic fibrosis can only be screened for up to eight weeks of age.

Please speak to your midwife, health visitor or GP if you would like your baby to be screened, or if you would like further information or talk about any concerns.

Yours sincerely,

Appendix 2: Blood transfusions – flowchart and scenarios



Scenario 1
A baby has had a blood transfusion on day 4, 5, 6 or 7. When should the ‘day 5’ sample be taken?

Solution
If the baby is transfused on day 4 the ‘day 5’ sample can be taken on day 8. If the baby is transfused on day 5, 6 or 7 the ‘day 5’ sample should ideally be taken before transfusion.

In the event of multiple blood transfusions, 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.
If the routine sample has been taken within 3 days (72 hours) of a transfusion, a repeat sample should be taken at least 3 days (72 hours) after the last transfusion.

Scenario 2
A baby completes its last blood transfusion at 11 am on 1 July. When should the repeat sample for CHT, CF and the IMDs be taken?

Solution
After 11am on 4 July. The repeat sample must be taken at least 72 hours after the last transfusion. This is to ensure the metabolite concentrations in the blood have returned to pre-transfusion levels.

Appendix 3: CHT preterm repeat – scenarios

Scenario 1
A baby is born at 32+0 weeks gestation. Is a CHT preterm repeat needed?

Solution
No – only babies born at less than 32 weeks (equal to or less than 31+6 weeks) should be offered a CHT preterm repeat.

Scenario 2
A baby is due to be discharged on day 27 and has a CHT preterm repeat taken. The baby then stays in hospital for another night. The laboratory asks for a repeat as the sample wasn't taken on the correct day.

Solution
If the baby is being discharged home before day 28, write **'discharged home'** on the blood spot card to ensure the laboratory knows why repeat sample was taken before day 28.

Scenario 3
A baby did not have a CHT preterm repeat taken on day 28 because he/she was given a blood transfusion on day 27 and was discharged home soon afterwards. The responsibility for taking the CHT preterm repeat was transferred to the community. The reason for this was to avoid the baby having a heel prick in hospital and a second heel prick 72 hours after the blood transfusion.

Solution
This scenario will occur infrequently. If a baby is fit for discharge but requires a top-up blood transfusion, treat as day of discharge and take the CHT preterm repeat sample pre-transfusion.

Scenario 4
A baby did not have a CHT preterm repeat screen at day 28 because he/she was given a blood transfusion on day 27 and then transferred to another neonatal unit.

Solution
Record clearly on the transfer documentation and IT system that screening is incomplete and transfer responsibility to complete screening to the receiving unit. Take the CHT repeat sample 72 hours after the last transfusion.

Scenario 5
A baby is born very prematurely. Should they have a CHT preterm repeat at day 28/discharge home AND when they reach 32 weeks gestation?

Solution
No – only one CHT preterm repeat is needed (at day 28 or day of discharge home, whichever is sooner).

Scenario 6
A baby is having multiple blood transfusions around day 28. Should a repeat be taken 72 hours after each transfusion?

Solution
No – only one repeat is needed, as soon as there is a 72 hour window. Ideally this should be as close to 28 days as possible.

Glossary

affected	In everyday speech, when someone has signs or symptoms of a condition, it is said that they are affected. However, in screening, affected is used to describe someone who has the condition, whether or not they might have signs or symptoms if untreated. In screening terms, a child who is affected with cystic fibrosis is a child who has the genetic make up for cystic fibrosis, whether or not they have signs or symptoms.
amino acid	Our bodies break down protein foods like meat and fish into amino acids (the building blocks of protein). Any amino acids that aren't needed are usually broken down and removed from the body. Babies with the inherited metabolic diseases that are screened for are unable to break down one or more amino acids. When levels of these amino acids get very high, they are harmful.
antenatal screening	Antenatal screening is screening which is carried out during pregnancy. This can include doing tests on the pregnant mother, her partner or the unborn baby. Antenatal screening includes tests for a wide range of conditions.
audit	A systematic comparison of screening, treatment and other management procedures with an agreed set of standards.
blood sampling	This refers to the collecting of blood to undertake tests. In the case of newborn screening it refers to the collection of small amounts of blood from the baby's heel. This is done by pricking the heel.
blood spot	A sample of blood that is taken from a baby's heel and spotted onto a special type of filter paper. A number of tests are then carried out on this blood spot. These tests are often called newborn blood spot screening.
blood transfusion	In the context of newborn screening, the transfusion of whole blood or any blood product that will affect the circulating concentration of the measured metabolite. The overall effect of any such transfusion will depend on a number of variables: • Circulating blood volume • Circulating concentration of metabolites • Distribution of metabolite between intracellular and extracellular compartments • Volume and rate of transfusion • Concentrations of metabolite in transfused fluid • Time since transfusion

	In practice, this refers to blood transfusions, exchange transfusions, platelets and fresh frozen plasma. An interval of at least 3 days (72 hours) is required between a transfusion of any of these and a blood spot sample. If a blood spot sample has been taken within 3 days (72 hours) of a transfusion, a repeat sample should be taken at least 3 days (72 hours) after the last transfusion. We recommend that albumin transfusions should **not** be included in the definition and that this should not delay the taking of the samples.
calcaneus	The bone of the heel.
child health records department (in the past, often referred to as 'child health')	The child health records department has records of all children in the area. When a mother gives birth the child health records department is notified of the birth. The results of newborn and other screening tests are also reported to child health records departments.
condition	There are lots of different words used to describe illnesses. They are sometimes called diseases, or disorders, or conditions.
congenital hypothyroidism	Babies with CHT do not have enough of the hormone thyroxine. Without thyroxine babies do not grow properly and can develop permanent, serious physical problems and learning disabilities. Babies with CHT can be treated early with thyroxine tablets and this will allow them to develop normally. CHT has been screened for throughout the UK since 1981.
consent	Agreement to a plan of action or particular treatment having received full information about the risks and benefits ('informed consent').
coverage	When talking about screening programmes, people often talk about coverage. This is the proportion of people actually screened. This is usually measured as a percentage. The success of screening programmes is sometimes measured by the coverage achieved.
cystic fibrosis	This inherited condition affects the digestion and lungs. Babies with CF may not gain weight well, have frequent chest infections and a limited life span. If babies with CF are treated early with a high-energy diet, medicines and physiotherapy, they may live longer, healthier lives. Screening for CF was introduced in England in 2007.

diagnosis / diagnostic test	A screening test distinguishes those at higher risk of a condition, from those at a lower risk. A diagnostic test is more definitive and can be used to confirm whether or not someone has a condition. Diagnostic tests often follow screening tests. For example a newborn baby might be screened for cystic fibrosis. The screening result shows that the baby probably has the condition. Further diagnostic tests will then be carried out to find out whether the child definitely has cystic fibrosis. This is then considered the confirmed result.
disease	There are lots of different words used to describe illnesses. They are sometimes called diseases, or disorders, or conditions.
disorder	There are lots of different words used to describe illnesses. They are sometimes called diseases, or disorders, or conditions.
false-negative result	A false-negative result is one where the person is thought not to have the condition, but then turns out to do so. For example, when a child who has a negative screening result for cystic fibrosis (and is therefore thought not to have the condition) turns out to have cystic fibrosis.
false-positive result	A false-positive result is one where the result is positive, but the person turns out not to have the condition. For example, when a child who has a positive screening result for CHT (and is therefore thought to be affected on the basis of the screening result) turns out not to have CHT. For parents, receiving a false-positive result can mean that they think that their child is sick, when actually they are healthy.
glassine envelope	Glassine is a light-weight, semi-transparent material that contains no chemicals which can harm the sample and is fairly resistant to moisture.
glutaric aciduria type 1	An inherited metabolic disease that prevents the break down the amino acids lysine and tryptophan contained within protein. For people with GA1, eating normal amounts of protein can cause harmful substances to build up in the blood and urine. In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Without treatment, the child can go into a coma. Though most children come out of the coma, they usually have brain damage that affects their ability to control their muscles and movements. This means that they may be unable to sit, walk, talk or swallow. GA1 can be treated with a protein-restricted diet and carnitine. A different regimen is required when the child is ill, and they may need to be hospitalised. Screening for GA1 was introduced in England in January 2015.
homocystinuria	An inherited metabolic disease that prevents the breakdown of the amino acid homocysteine contained within protein. This then builds up in the blood. In the long term, this can lead to a number of health

	problems. Without treatment, most children with HCU have learning difficulties and eye problems. They may also develop bones that are abnormally long and thin (osteoporosis), and blood clots or strokes. HCU can be treated with a protein-restricted diet and extra supplements and medicines. Screening for HCU was introduced in England in January 2015.
inherited metabolic disease	A genetic disease that affects the metabolism. Babies with inherited metabolic conditions cannot process certain substances in their food. Without treatment babies with some of these conditions can become suddenly and seriously ill. The symptoms of the conditions are different; some may be life threatening or lead to severe developmental problems. They can all be treated by a carefully managed diet, which is different for each condition and may include additional medicines.
isovaleric acidaemia	An inherited metabolic disease that prevents the breakdown of the amino acid leucine contained within protein. For people with IVA, eating normal amounts of protein can cause harmful substances to build up in the blood. Children with IVA can become severely unwell. Without treatment, this can lead to a coma and permanent brain damage. Some babies with IVA have problems within a few days of birth; other children become unwell at a few months or years of age, maybe during a minor illness, such as a chest infection or a tummy upset. IVA can be treated with a protein-restricted diet and carnitine and glycine. A different regimen is required when the child is ill, and they may need to be hospitalised. IVA can vary in severity. In some mild forms of IVA, the risk of problems is much lower and this means that the treatment can be simpler. Screening for IVA was introduced in England in January 2015.
manual lancet	A lancet that is pushed into the tissues by hand. It does not allow for accurate control of the depth of the puncture.
maple syrup urine disease	An inherited metabolic disease that prevents the breakdown of the amino acids leucine, isoleucine and valine contained within protein. For people with MSUD, eating normal amounts of protein can cause a harmful build-up of these amino acids in the blood. Many babies with MSUD become unwell when they are a few days old. Without treatment, this leads to a coma and permanent brain damage. In older children a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. As in babies, this can lead to a coma unless treated correctly. MSUD can be treated with a protein-restricted diet. A different regime is

	required when the child is ill, and they may need to be hospitalised. Screening for MSUD was introduced in England in January 2015.
medium-chain acyl-CoA dehydrogenase deficiency	An inherited metabolic disease belonging to a group known as fatty acid oxidation disorders where there is a deficiency of a mitochondrial enzyme. This makes it difficult for the body to break down fatty acids and produce energy, and can cause sudden death in infants. Most of the time children are well, but an infection or relatively long period without food upsets their metabolism causing coma and sometimes death. Treatment involves ensuring that children do not go for long periods without food and special management if they do get an infection. Periods of not eating can safely get longer as the child grows. Screening for MCADD was introduced in England in 2009.
newborn screening	All screening on a newborn baby is called newborn (or neonatal) screening. Current newborn screening includes hearing screening, screening for abnormal; hips and other physical problems, and blood spot screening.
normal (result)	Sometimes when the result of the test shows that the child is unlikely to have the condition tested for, people say the result is normal.
personal child health record	This is the child health record which is held by the parent, also called the ‘red book’. It is normally issued by the midwife or health visitor.
phenylketonuria	An inherited metabolic disease that prevents the breakdown of the amino acid phenylalanine contained within protein. For people with PKU, eating normal amounts of protein can cause a harmful build-up of this amino acid. If left untreated this leads to poor brain development. If identified early the child can be put on a restricted-protein diet with supplements and the brain can develop normally. PKU has been screened for throughout the UK since 1969.
screen-negative result	Screening results are not 100% conclusive. Instead they provide presumptive results. A screen-negative result is a result which suggests that the child does not have the condition for which they are being screened. Sometimes people will say that the result is “normal”. A screen-negative result for CF means that it is highly likely that the child does NOT have CF. This screen-negative result is NOT usually confirmed using further tests, but it is assumed the child is not affected.
screen-positive result	Screening results are not 100% conclusive. Instead they provide presumptive results. A screen-positive result is a result which shows

	that the child is likely to have the condition for which they are screened. Sometimes people will say that the child is affected. Positive screening results are then confirmed using diagnostic tests.
screening	Screening is when healthy children and adults are tested to see if they are likely to develop a condition. Screening tests don't generally confirm that a person has a disease. Usually they will not feel ill from these conditions in any way at the time when they're screened. Screening allows diseases to be identified early, before any signs of illness. This means people can be treated quickly and hopefully avoid getting seriously ill. Screening happens at different ages, and for different conditions. Newborn blood spot screening in England includes tests for sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT) and six inherited metabolic diseases (IMDs): phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive) (HCU).
sickle cell disease	Sickle cell disease affects haemoglobin, a part of the blood that carries oxygen around the body. Babies who have these conditions will need specialist care throughout their lives. People with SCD can have attacks of severe pain, get serious, life-threatening infections and are usually anaemic (their bodies have difficulty carrying oxygen). Babies with sickle cell disease can receive early treatment, including immunisations and antibiotics, which, along with support from their parents, will help reduce the chance of serious illness and allow the child to live a healthier life. Screening for SCD was introduced in England in 2001.
UK National Screening Committee	This is a national advisory body which makes recommendations about screening to the UK Departments of Health.
NHS Newborn Blood Spot Screening Programme	This refers to the national programme in England that works in partnership with those organising newborn blood spot screening locally to support a high quality service responsive to the needs of families. The English programme also works in partnership with the blood spot screening programmes in Scotland, Wales and Northern Ireland to deliver a high quality service across the UK.

Abbreviations

CF	cystic fibrosis
CHRD	child health records department
CHT	congenital hypothyroidism
DNA	deoxyribonucleic acid
EDTA	ethylenediaminetetraacetic acid
GA1	glutaric aciduria type 1
GP	general practitioner
HCU	homocystinuria
IMD	inherited metabolic disease
IRT	immunoreactive trypsinogen
IT	information technology
IVA	isovaleric acidaemia
KPI	key performance indicator
MCADD	medium-chain acyl-CoA dehydrogenase deficiency
MSUD	maple syrup urine disease
NBS	newborn blood spot
NBSFS	Newborn Blood Spot Failsafe Solution
NHS	National Health Service
NMC	Nursing and Midwifery Council
PCHR	personal child health record (“red book”)
PHE	Public Health England
PKU	phenylketonuria
SCD	sickle cell disease
TSH	thyroid stimulating hormone
UK NSC	UK National Screening Committee