Newborn Bloodspot Screening

See also: Competency - To correctly take a newborn blood spot test.

Purpose
Universal screening of all infants at 5 days of age for
  1. phenylketonuria (PKU)
  2. congenital hypothyroidism (CHT)
  3. sickle cell disease (SCD)
  4. cystic fibrosis (CF)
  5. medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

Treatment for PKU and CHT should commence by the time the baby is 21 days old. Guidance from the UK Newborn Screening Programme Centre means that a baby may require more than one blood spot test. It is not necessary for a baby to be on feeds before a sample can be taken.

Key Points

- Day 0 is the date of birth regardless of the time born. The aim is that all newborn babies should have a blood spot test taken on Day 5.
- Babies transfused before Day 5 are tested before and >72 hours after transfusion.
- Tests must be performed by Day 8 at the latest, even if multiple transfusions are ongoing, to ensure timely detection and treatment of PKU
- Babies born <35/40 will require a repeat blood spot at 37/40 (term) corrected gestational age
- To minimise discomfort, tests should be coordinated with other investigations that the baby requires for his/her clinical management (e.g. blood sugars etc)
- Fill the cards in completely including the NHS number as baby’s name may change
When to perform the blood spot testing

Admission (Day 0)  If the baby is less than 1000g, or is potentially likely to have a blood transfusion before Day 5, take a single circle blood spot on admission, prior to transfusion. Mark it “pre-transfusion” and keep it in the pocket in the front of the notes (see Day 5, below).

This sample is for sickle cell disease (SCD) screening. It is anticipated that only a small number of babies each year (typically the extremely preterm, or the term baby requiring full exchange transfusion) will fall into this category. Blood taking should be coordinated with clinical testing wherever possible.

Day 5  Take a full (4 circle) blood spot test on all babies

Complete all boxes on the card including the NHS number. If the baby has had a blood transfusion then the blood spot should be delayed until >72 hours post-transfusion (i.e. Day 5 to Day 8). In the event of multiple transfusions, a full blood spot must be sent by Day 8 regardless.

Record the date of transfusion, tick the ‘repeat’ box and send this card stapled together with the pre-transfusion blood spot card. If the baby was not transfused before the Day 5 sample, then the initial single spot should be discarded and not sent to the screening laboratory.

Preterm  When a preterm infant (born <35/40) reaches 36 weeks equivalent, a single circle repeat blood spot is required for CHT. This is because immaturity can mask congenital hypothyroidism. Give gestational information on form. Babies born >35/40 should have a four-spot test performed on Day 5, and do not require a repeat unless requested by the screening laboratory.

Discharge  Blood spot status (what taken and when) should be recorded on Badgernet so that it is included in the discharge summary. The summary must include details of any repeat samples that are needed and liaison with community team.

The laboratory may request repeat screening in the event of discrepancies or abnormalities being detected.

Sending samples

Samples should arrive in the screening laboratory (St. Helier Hospital) within 2-4 working days. Cards will be collected by the Community Midwives clerk on Joan Booker ward. They will send them off on the next working day, first thing in the morning.

Arrangements must be made with the community neonatal team to collect repeat samples (transfused infants or preterm infants at 36/40) as there are no reminders sent from the lab.

The laboratory will request further samples if there are discrepancies in the sample dates, or if there are abnormalities in the test results. These should be considered to be urgent.

Samples should be sent within 24 hours of being taken to reach the lab in 48 hours. Samples taken at the weekend or on bank holidays are sent on the next working day.

For further information see http://newbornbloodspot.screening.nhs.uk/

In the event of technical or administrative enquiries, please contact the Community Midwives office (extension 2413) or the Regional Newborn Screening Service in the Department of Chemical Pathology at St Helier Hospital on 020 8296 2991.
At or after 72 hrs post-transfusion repeat the blood spot screening (Day 5 to Day 8). Do not delay later than Day 8 regardless (e.g. if multiple transfusions).

Blood spot normally performed by midwifery staff if = Day 5 but check as may have been delayed.

Day 0-4 requires blood transfusion

Yes

No

Day 5 Full Blood spot test to be taken

If pre-transfusion spot not taken, then book repeat for 3 months after last transfusion

At corrected age of 36 weeks repeat single blood spot screening. If baby home before then liaise with community nursing team

Born at less than 36 weeks gestation?

No repeats required unless requested by screening laboratory

Yes

Admitted to neonatal unit within 5 days of birth

No
## How to take Blood Spot Screening at St. Peter’s Hospital

### Equipment
- Parent information leaflet (should be given to parents – see Appendix A to download)
- Non-sterile protective gloves
- Microtainer lancet / Unistik
- Cotton wool ball
- Plaster
- Blood spot card and glassine envelope
- Blood spot book (NICU only)
- Nursing and medical records

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Rationale</th>
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<tr>
<td>Can the sample be done at the same time as other blood tests? – communicate!</td>
<td>To minimise discomfort and handling</td>
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<tr>
<td>Explain to parents why the newborn blood spot screening test is performed and how it is taken. Give information leaflet (can download from Appendix A) if parents don’t already have it.</td>
<td>To obtain parental understanding and verbal consent. Record decision to screen in notes.</td>
</tr>
<tr>
<td>Fill in all the boxes on the newborn blood spot card, confirm baby’s name, date of birth and parent’s contact details.</td>
<td>To identify patient with results.</td>
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<tr>
<td>Wash hands and put on non-sterile protective gloves.</td>
<td>To prevent cross infection</td>
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<tr>
<td>The heel should be washed with plain tap water. Ensure the baby’s heel is clean and dry.</td>
<td>Washing is important for test reliability. To prevent contamination of the sample or infection of the puncture site.</td>
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<tr>
<td>Using the lancet on the inner or outer plantar aspects of the heel, gently pierce the skin.</td>
<td>To minimise skin trauma and maximise potential to obtain blood. Topical pain relief cannot be given as this may contaminate the sample.</td>
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<tr>
<td>Wait up to 15 seconds to allow blood to flow. Apply the blood drop to one side of the card. Allow the blood to fill the circle by natural flow, and seep through to the back of card. Fill all the circles completely and avoid layering blood.</td>
<td>Less than four circles is inadequate, leads to inaccurate results and greater likelihood of a repeat procedure. The exception to this is the pre-transfusion sample, for which a single blood spot is sufficient. The term repeat sample should be a four circle sample.</td>
</tr>
<tr>
<td>Apply cotton wool to the wound, then apply a plaster to the heel if appropriate / required.</td>
<td>To stop bleeding.</td>
</tr>
<tr>
<td>Dispose of the sharp in a sharps container.</td>
<td>To prevent injury and cross infection to others</td>
</tr>
<tr>
<td>Allow blood spots to air-dry and put the card in the envelope.</td>
<td>To prevent sample contamination/damage</td>
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<tr>
<td><strong>On NICU/TCR</strong>, put the completed card into the blood spot box for collection.</td>
<td>Sample card will be collected by clerk.</td>
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<tr>
<td>Record in the blood spot book that the blood spot has been performed, with the date taken and sign next to the correct baby’s name. State if it is a repeat test.</td>
<td>To keep accurate records, giving full information about transfusions, previous blood spot cards etc</td>
</tr>
<tr>
<td><strong>Postnatal ward / Community / Paediatric Wards</strong> send / bring the completed blood spot card to the blood spot coordinator Community Midwives Office, Joan Booker Ward.</td>
<td>To ensure test is sent to appropriate place for analysis.</td>
</tr>
<tr>
<td>Record when blood spot was performed on the nursing charts, and the nursing and medical records.</td>
<td>To keep accurate records.</td>
</tr>
<tr>
<td>If the baby has had a blood transfusion and no pre-transfusion card was taken, make a note that the test needs to be repeated at or after 72 hours for PKU, CHT, (CF) and again at 3 months for SCD.</td>
<td>To ensure test for Phenylketonuria is not missed and to ensure that haemoglobinopathy screening is performed correctly. A risk management form should be filled in.</td>
</tr>
<tr>
<td>When the repeat test is taken, record it as above.</td>
<td>To keep accurate records.</td>
</tr>
<tr>
<td>Record any subsequent tests as above</td>
<td>To keep accurate records.</td>
</tr>
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Some key facts for staff to assist in talking to parents

Why do we screen babies?
1 Explain that screening is designed to identify babies at highest risk of certain rare conditions.
2 Specify the conditions screened for in your area.
3 Explain that screening aims to identify babies with these conditions BEFORE they develop symptoms so that they can be started on treatment quickly.
4 Explain that early treatment can then improve the health of babies with these rare conditions, for babies with PKU and CHT severe mental disability can be prevented.
5 Explain that although screening can be an anxious process for families, it does prevent the anxiety associated with uncertainty over symptoms before a clinical diagnosis is made.

What can screening achieve?
1 Explain that whilst screening is positively encouraged, parents have the right to choose.
2 Stress that screening is not the same as diagnosis. It is only the first step, identifying babies at highest risk of having the conditions. These babies will need further tests before a diagnosis can be made.
3 Explain that, very rarely, babies with the conditions will be missed (false-negatives); and, very rarely, babies without the conditions will be wrongly identified (false-positives).
4 Stress that even where the screening tests themselves are 'perfect', screening will 'fail' if parents are not told the results and babies are not started on treatment.

Phenylketonia (PKU)
1 Explain that PKU affects 1 in 10,000 babies across the UK.
2 Explain that whilst this is an inherited condition, carriers are not identified as part of the screening. Parents of babies who are found to be affected by PKU must, by definition, both be carriers and will be offered counselling regarding the risk to future pregnancies.
3 Because babies with PKU cannot digest phenylalanine, an amino acid present in many of the foods we eat, their brains are unable to develop properly.
4 Stress that this means that the impact on a baby’s brain development is more serious the longer they go untreated. If a baby is never treated they develop serious, irreversible, mental disability.
5 If babies are started on treatment by 21 days of age this disability can be prevented.

Congenital Hypothyroidism (CHT)
1 CHT affects 1 in 4,000 babies across the UK.
2 There are several different causes of congenital hypothyroidism, and only 1 in 10 cases are inherited. Carriers are not identified as part of the screening. Parents of babies with CHT may want access to tests and counselling to inform their decisions about future pregnancies.
3 Because babies with CHT do not have enough thyroxine, a hormone necessary for growth, their brains are unable to develop properly.
4 Stress that this means that the impact on a baby’s brain development is more serious the longer they go untreated. If a baby is never treated they develop serious, irreversible, mental disability.
5 If babies are started on treatment by 21 days of age this disability can be prevented.

Sickle cell disorders (SCD)
1 About 1 in 2,500 babies in the UK are affected with sickle cell disorders.
2 Sickle cell disorders are inherited. The screening process for SCD also identifies carriers. Parents of babies with SCD, and babies who are carriers, may want access to counselling regarding the risk to future pregnancies.
3 Sickle cell disorders cause red blood cells to become sickle-shaped, and that this can cause pain, tissue damage, infection and even death.
4 Early treatment by 2 months of age can improve the health of babies with SCD and prevent deaths.
Cystic fibrosis (CF)

1. Explain that CF affects 1 in 2,500 babies across the UK.
2. Explain that CF is an inherited condition. Parents of babies with CF may want access to counselling regarding the risk to future pregnancies.
3. The screening process for CF includes testing some babies’ DNA for the most common CF-causing gene alterations; therefore some carriers of CF will be identified. Carrier results will be reported to parents. For some babies identified as carriers, CF cannot be ruled out; some parents may want access to further tests and/or genetic counselling to discuss these risks.
4. Explain that CF affects babies’ digestion and lungs, and babies fail to thrive.
5. Explain that screening is thought to improve the outlook for babies with CF, and to improve parents’ experience of diagnosis. Without screening, children can be ill for some time before a diagnosis is made.
6. Mention the treatments available, and explain that the symptoms, severity and treatment for CF can be very varied.

Consultation about re-testing >36 weeks corrected age

Review of the current policy for repeat testing of preterm infants has been ‘fast-tracked’ ahead of the review of CHT screening policy planned for 2011 and which includes screening and diagnostic protocols and other aspects contained within the initial clinical referral guidelines. The consultation period lasted for 4 weeks and has now closed.

Please note that TSH cut-offs used currently to define presumptive positive and borderline results will continue until the review of CHT screening policy is concluded in 2011.

References

Policies and Standards for Newborn Blood Spot Screening April 2005
Green A: Neonatal Screening: current trends and quality control in the United Kingdom; Rinsho Byori. 46(3):211-6, 1998

Guideline

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Appendix A

All information is now found at http://newbornbloodspot.screening.nhs.uk/

The current leaflets are rather large (32 or 72 pages) and are also available in other languages.