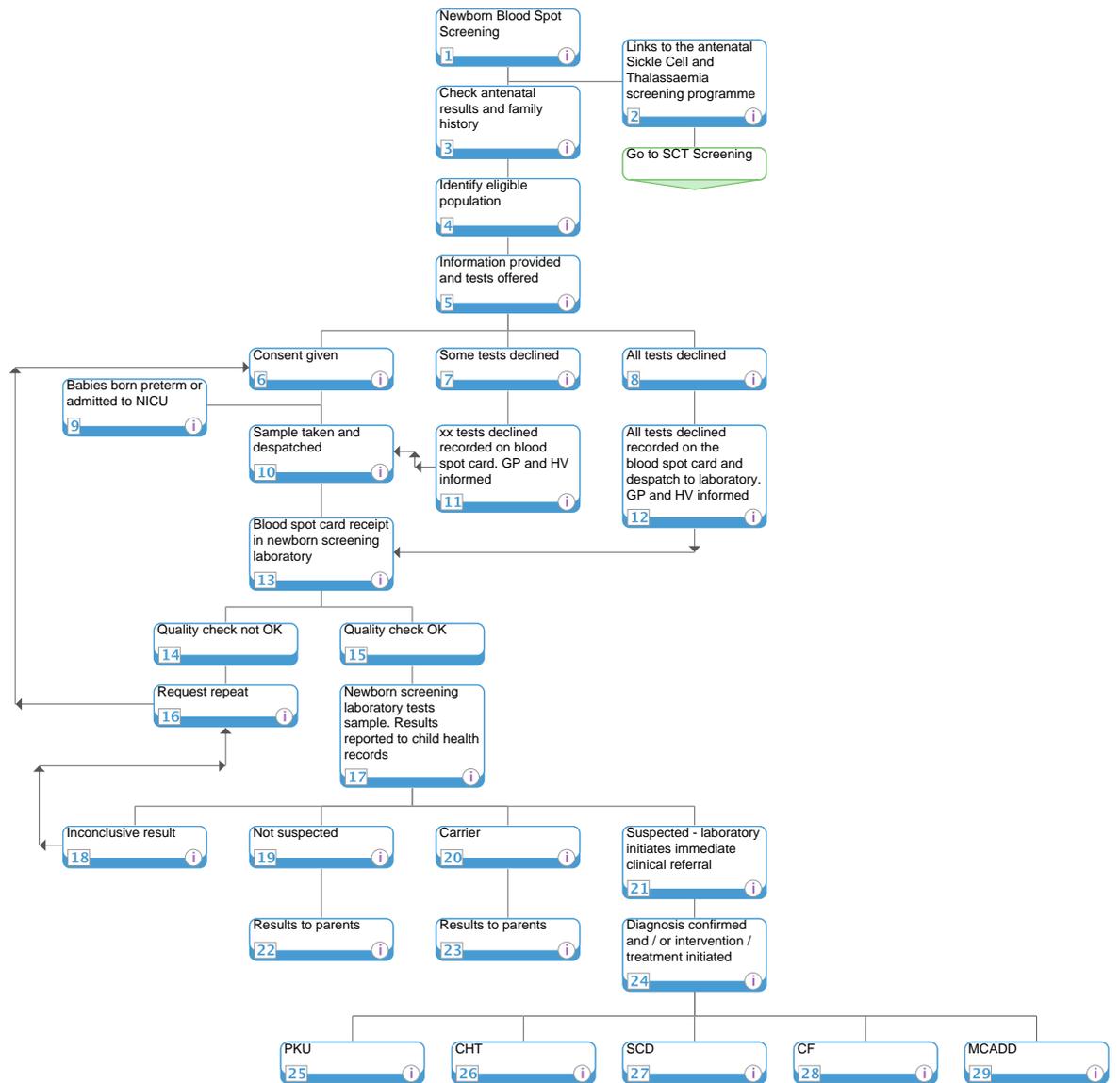


Newborn Blood Spot Screening

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1 Newborn Blood Spot Screening

Quick info:

Newborn blood spot screening (NBS) is a routine test, offered to all parents, for babies between 5 and 8 days of age (ideally at day 5 counting date of birth as day 0).

The baby's heel is pricked using a special lancet device and some drops of blood collected onto a blood spot card.

The blood is tested to identify babies at high risk of five rare conditions:

- Phenylketonuria (PKU) 1:10,000
- Congenital Hypothyroidism (CHT) 1: 4,000
- Sickle Cell Disease (SCD) 1:1,900
- Cystic Fibrosis (CF) 1:2,500
- Medium Chain acyl CoA Dehydrogenase Deficiency (MCADD) 1:10,000-20,000

The overall goal is for early detection and referral of babies, found to be high risk, to improve health and prevent severe disability or even death.

<http://newbornbloodspot.screening.nhs.uk/>

2 Links to the antenatal Sickle Cell and Thalassaemia screening programme

Quick info:

For parents, the link between antenatal and newborn screening is obvious: the result of an antenatal genetic test will be related to the results of a newborn baby's genetic test for the same condition, so the linked programme provides a natural failsafe check between the mother and baby result.

The antenatal report given to women should contain all antenatal screening results for communication to primary care and allow linkage to newborn screening results where relevant.

There should be systems in place to inform newborn screening laboratories of all antenatal screening and diagnostic results and there should be a named person in every maternity unit with responsibility to ensure that newborn screening laboratories are informed of carrier women whose pregnancy is ongoing.

More information is available at <http://sct.screening.nhs.uk/linkage>

3 Check antenatal results and family history

Quick info:

Antenatal sickle cell and thalassaemia screening results should be recorded on the blood spot card.

Infants with a family history of a metabolic disorder may need special testing in the neonatal period and should be referred to a paediatrician in the antenatal period.

4 Identify eligible population

Quick info:

The eligible population includes babies up to one year of age.

The population is identified by:

- NN4B issued at birth notification
- NN4B issued at registration with a GP practice for babies born abroad that were not issued with an NHS number at birth.

Recorded on the child health records department (CHRD) IT system in the responsible PCT.

5 Information provided and tests offered

Quick info:

Pregnant women are given the UK NSC information booklet 'Screening tests for you and your baby' at their booking appointment and newborn screening is discussed towards the end of pregnancy and at least 24 hours pre-test.

Antenatal sickle cell and thalassaemia screening should be offered to all women and couples in pregnancy by 8-10 weeks to allow for the option of prenatal diagnosis.

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The booklet 'screening tests for your baby' can be issued if parents have not kept or did not receive the UK NSC booklet earlier (e.g. families of babies born abroad). This contains newborn screening information only. Both booklets and translated copies are available at <http://www.screening.nhs.uk/annbpublications>

6 Consent given

Quick info:

Consent and reference to the provision of written information must be recorded in the maternity/baby health records and Personal Child Health Record (sometimes referred to as the 'Red book').

7 Some tests declined

Quick info:

Decline and reference to the provision of written information must be recorded in the maternity/baby records and Personal Child Health Record. Inform parents who to contact if they change their minds.

Communicate parents' wishes to laboratory by writing which condition(s) have been declined on the blood spot card.

Inform the GP and health visitor, which tests, have been omitted so they are aware, should symptoms arise, that the possibility of an affected child cannot be ruled out.

Send the blood spot card to the newborn screening laboratory. The child health records department is notified of the parents' decision to decline by the normal communication lines via the laboratory.

8 All tests declined

Quick info:

Decline and reference to the provision of written information must be recorded in the maternity/baby records and Personal Child Health Record. Inform parents who to contact if they change their minds.

Communicate parents' wishes to laboratory by writing blood spot screening declined on the blood spot card.

Inform the GP and health visitor, which tests, have been omitted so they are aware, should symptoms arise, that the possibility of an affected child cannot be ruled out.

Send the blood spot card to the newborn screening laboratory. The child health records department is notified of the parents' decision to decline by the normal communication lines via the laboratory.

9 Babies born preterm or admitted to NICU

Quick info:

Babies admitted to neonatal intensive care units at less than 5 days of age should have a single circle blood spot sample taken and marked as 'PRE-TRANSFUSION'

Please check that the pre-transfusion blood spot card has been taken and sent to the laboratory in accordance with local pathway.

Where a baby has already had a blood transfusion, either intrauterine or in the newborn period, before the screening blood sample has been taken, repeat samples are needed at 72 hours (3 days) after the blood transfusion for phenylketonuria, congenital hypothyroidism, cystic fibrosis and Medium Chain acyl CoA Dehydrogenase Deficiency (MCADD)

Where a baby has already had a blood transfusion, either intrauterine or in the newborn period, before the screening blood sample has been taken for sickle cell, the sample is sent for DNA analysis. This test identifies babies who might have sickle cell disease or babies who are sickle cell carriers, but not some of the rarer haemoglobin disorders or carriers of other haemoglobin variants identified on the routine test for sickle cell disease.

Repeat blood spot samples should be taken in preterm babies for congenital hypothyroidism (this policy is under review please refer to <http://newbornbloodspot.screening.nhs.uk/cht>)

Babies admitted to neonatal intensive care units are likely to have multiple blood samples taken. Blood spot screening should be coordinated with other tests. Venepuncture or venous/arterial sampling from an existing line is an alternative, this is providing the sample is not contaminated with heparin and the line cleared of infusate.

Analgesia in the form of breast feeding, non-nutritive sucking and a dose of sucrose or glucose is recommended for babies who undergo multiple invasive procedures.

Full guidelines available at <http://newbornbloodspot.screening.nhs.uk/bloodspotsampling>

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10 Sample taken and despatched

Quick info:

The sample should be taken in accordance with Guidelines for Newborn Blood Spot Sampling <http://newbornbloodspot.screening.nhs.uk/bloodspotsampling> and despatched within 24 hours (preferably same day), in an identifiable screening envelope and by first class post.

The baby's NHS number must be recorded on the blood spot card.

Record sickle cell and thalassaemia parental antenatal screening results on the blood spot card.

An online map of newborn screening laboratories and maternity service is being developed and will be available in early 2011.

11 xx tests declined recorded on blood spot card. GP and HV informed

Quick info:

See the information in the 'some tests declined' box, above.

12 All tests declined recorded on the blood spot card and despatch to laboratory. GP and HV informed

Quick info:

See the information in the 'all tests declined' box, above..

13 Blood spot card receipt in newborn screening laboratory

Quick info:

The newborn screening laboratory checks that the sample is satisfactory and that there is sufficient blood to complete screening on all five tests and that all data fields are completed to enable:

- The baby to be identified (NHS number is mandatory and bar-coded labels preferred)
- The laboratory to analyse the blood (including antenatal sickle cell and thalassaemia screening results)
- Identify the responsible PCT to report the results.

Sample receipt in the laboratory is notified to child health records and some maternity units, using the status code 01 to assist the timely identification of untested babies.

16 Request repeat

Quick info:

A repeat test may be requested because:

- Results are uncertain
- The sample is not valid (e.g. taken too early)
- There is insufficient blood

There is inadequate data to

- Identify the baby
- Analyse the sample
- Report the results

A local policy for repeat testing of babies must be in place, reasons for repeat tests must be communicated to the midwife in writing and passed onto parents.

17 Newborn screening laboratory tests sample. Results reported to child health records

Quick info:

Screening test results should be communicated to the child health records departments daily, using the screening status codes

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(To access screening status codes <http://newbornbloodspot.screening.nhs.uk/getdata.php?id=11004>)

18 Inconclusive result

Quick info:

Results that are inconclusive require an additional sample to be obtained to complete the screening test. e.g. this may occur in cystic fibrosis and congenital hypothyroidism when the result is borderline

Information on repeat test for CF can be found on http://newbornbloodspot.screening.nhs.uk/hp_comms_guidelines

19 Not suspected

Quick info:

The term 'not suspected' is used because screening test are not 100% certain.

Screening identifies babies who are genetic carriers of sickle cell or other unusual red blood disorders. Carriers of sickle cell are usually healthy and not affected by the condition (see the information in the 'results to parents' box, below).

Screening for CF includes testing some babies for the most common gene changes that cause CF. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have CF (see the information in the 'results to parents' box, under the 'carrier' box to the right).

20 Carrier

Quick info:

Carriers of cystic fibrosis

Screening for cystic fibrosis includes testing some babies for the most common gene changes that cause cystic fibrosis. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have cystic fibrosis (see the information in the 'results to parents' box, below).

Carriers of Sickle cell

Carriers of Sickle cell are usually healthy and not affected by the condition (see the information in the 'results to parents' box under the 'not suspected' box).

Carriers of MCADD are not detected by the screening test but may be detected through diagnostic testing.

21 Suspected - laboratory initiates immediate clinical referral

Quick info:

The laboratory reports phenylketonuria, congenital hypothyroidism, cystic fibrosis and MCADD screen positive results directly to the paediatric specialist team by telephone and in writing, the GP is also informed in writing.

Communication flows and guidelines for clinical referral of each of the conditions are available

- phenylketonuria (PKU) <http://newbornbloodspot.screening.nhs.uk/pku>
- congenital hypothyroidism (CHT) <http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10941>; pages 27-29 (currently under review)
- cystic fibrosis (CF) <http://newbornbloodspot.screening.nhs.uk/cf-resources>
- MCADD <http://newbornbloodspot.screening.nhs.uk/mcadd>
- SCD <http://sct.screening.nhs.uk/getdata.php?id=10763>; the laboratory reports screen positive sickle cell results according to locally agreed pathway

22 Results to parents

Quick info:

The child health records department notifies results to health visitors for reporting to parents.

Best practice is for the child health records department to inform parents of normal results by letter.

The screening results are recorded in the personal child health record.

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Results to parents, carriers of sickle cell

Robust processes should be in place to report newborn screening carrier results to parents. The format for reporting should be agreed locally but could be a face-to-face meeting or letter as appropriate.

For sickle cell screening carrier results, these should routinely be linked to maternal (and paternal) results where these are available.

23 Results to parents

Quick info:

Results to parents, carriers of cystic fibrosis

All women and their partners with an infant who has a cystic fibrosis carrier result should be informed of the result, receive relevant information and material about the result, and be offered access to an appropriately trained healthcare professional to discuss the result. GPs should also routinely receive information about the baby's carrier result.

A trained health care professional will make a visit to the family to inform them that their baby is thought to be a carrier of cystic fibrosis. Communication guidelines to support health professionals can be found at http://newbornbloodspot.screening.nhs.uk/hp_comms_guidelines

The cystic fibrosis carrier leaflet should be given <http://newbornbloodspot.screening.nhs.uk/cms.php?folder=2461>

24 Diagnosis confirmed and / or intervention / treatment initiated

Quick info:

The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome. Confirmatory tests and initial management of the conditions can be found at:

- phenylketonuria (PKU) <http://newbornbloodspot.screening.nhs.uk/pku>
- congenital hypothyroidism (CHT) <http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10941>; pages 27-29 (currently under review)
- cystic fibrosis (CF) <http://newbornbloodspot.screening.nhs.uk/cf-resources>
- MCADD <http://newbornbloodspot.screening.nhs.uk/mcadd>
- sickle cell (SCD) <http://sct.screening.nhs.uk/getdata.php?id=10763>; parents should be notified of screen positive sickle cell results by 4 weeks of age of the baby, and affected babies should attend local clinic by 3 months of age. Prophylactic penicillin is offered by 3 months of age to reduce associated symptoms and risk. <http://sct.screening.nhs.uk/getdata.php?id=10628>

25 PKU

Quick info:

The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome. Confirmatory tests and initial management of PKU can be found at:

PKU <http://newbornbloodspot.screening.nhs.uk/pku>

26 CHT

Quick info:

The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome. Confirmatory tests and initial management of CHT can be found at: <http://newbornbloodspot.screening.nhs.uk/getdata.php?id=10941>; pages 27-29 (currently under review)

27 SCD

Quick info:

Confirmatory tests can be found at: <http://sct.screening.nhs.uk/getdata.php?id=10756>

Confirmatory tests should be processed by laboratories who have experience in handling neonatal blood samples.

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Sickle cell centres and local hospitals should have robust follow up arrangements to identify and follow up any child who does not attend their hospital appointments and should be able to track children who have moved out of the area in order to make appropriate handover arrangements.

Infants should be prescribed oral penicillin by 3 months of age, routine Prevenar vaccinations and additional pneumococcal prophylaxis as outlined in the standards by 15 months of age.

Additional clinical guidelines can be found in the programme standards:

<http://sct.screening.nhs.uk/getdata.php?id=10763>

<http://sct.screening.nhs.uk/getdata.php?id=10628>

28 CF

Quick info:

The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome. Confirmatory tests and initial management of CF can be found at: <http://newbornbloodspot.screening.nhs.uk/cf-resources>

29 MCADD

Quick info:

The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome. Confirmatory tests and initial management of MCADD can be found at: <http://newbornbloodspot.screening.nhs.uk/mcadd>. Carrier status may be identified.

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Key Dates

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Evidence summary for Newborn Blood Spot Screening

References

This is a list of all the references that have passed critical appraisal for use in the pathway Newborn Blood Spot Screening

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