Healthcare Improvement Scotland

Pregnancy and Newborn Screening:

Newborn Bloodspot Standards

January 2019

NHS SCOTLAND

We are committed to equality and diversity. These standards are intended to support improvements in healthcare for everyone, regardless of their age, disability, gender reassignment, marriage and civil partnership, pregnancy and maternity, race, religion or belief, sex, sexual orientation, socio-economic status or any other status. Suggested aspects to consider and recommended practice throughout the standards should be interpreted as being inclusive of everyone living in Scotland. We have assessed these standards for likely impact on the nine equality protected characteristics as stated in the Equality Act 2010. A copy of the impact assessment is available on request from the Healthcare Improvement Scotland Equality and Diversity Advisor.

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Introduction

Background to the pregnancy and newborn screening standards

Screening is offered to groups of the population to identify people who may be at an increased chance of a particular condition. Pregnancy screening is offered to help women make informed choices about their health and the health of their unborn baby during pregnancy. Newborn screening is offered to facilitate parents and carers to make informed choices about the health of their newborn baby. It is important that informed decisions are made in partnership with healthcare professionals at the respective stage of the screening programme.[1] Information on pregnancy and newborn screening, including national information produced by NHS Health Scotland, is provided to women throughout their pregnancy and following the birth of the baby.

Newborn bloodspot screening

All newborn babies are offered a newborn bloodspot screening test between 96–120 hours (4–5 days).[2-5] The test is done by taking a blood sample from the baby's heel to determine if there are any health issues. The results highlight whether the baby is unaffected, is a healthy carrier, or has a condition.[6]

It is important to identify specific conditions as soon after birth as possible and before clinical symptoms begin.[7] Early identification will improve health outcomes and manage treatment options for the newborn baby.[7]

Policy context

National Services Division (NSD) has an ongoing role in the national multidisciplinary groups which monitor and evaluate the pregnancy and newborn screening programmes. Healthcare Improvement Scotland supports NHSScotland's screening programmes through developing new and, where appropriate, revising existing standards. A request to revise the *Clinical Standards for Pregnancy and Newborn Screening* (October 2005) was received from the Scottish Government and NSD in summer 2016.

NSD has developed and published a set of Key Performance Indicators (KPIs)[8] to document progress in areas that have been identified within the pregnancy and newborn screening programme as requiring additional support.

All the pregnancy and newborn screening standards should be read alongside other relevant legislation, standards and guidance such as the KPIs mentioned above.

Scope of the standards

These standards apply to newborn bloodspot screening and cover:

- eligibility and coverage
- sampling
- second sampling for cystic fibrosis and congenital hypothyroidism, and
- referral.

These newborn bloodspot screening standards should be read alongside the general standards for pregnancy and newborn screening:

- **Standard 1: Leadership and governance**
- **Standard 2: Information and support**

Information relating to the development of the pregnancy and newborn screening standards can be found in Appendix 1 of the general standards.

The full suite of pregnancy and newborn screening standards is available on the Healthcare Improvement Scotland website (www.healthcareimprovementscotland.org).

Format of the standards

All our standards follow the same format. Each standard includes:

- a statement of the level of performance to be achieved
- a rationale providing reasons why the standard is considered important
- a list of criteria describing the required structures, processes and outcomes
- what to expect if you are a person receiving care
- what to expect if you are a member of staff, and
- what the standards mean for organisations, including examples of evidence of achievement.

Within the standards, all criteria are considered 'essential' or 'required' in order to demonstrate the standard has been met.

Terminology

Wherever possible, we have incorporated generic terminology which can be applied across all health and social care settings.

The terms 'woman', 'women' and 'individual' are used within the standards to refer to all individuals with a female Community Health Index (CHI) number.

The term 'eligible women' refers to women who are invited for pregnancy screening.

The term 'parents and carers' is used within the standards to refer to the parent, caregiver or guardian who assumes legal parental responsibility for the newborn.

Quality of care approach

The pregnancy and newborn screening standards are a key component in supporting the pregnancy and newborn screening programme in quality assurance of its services. Monitoring and improving performance against these standards, at a local and national level, aims to improve the quality of the pregnancy and newborn screening programme.

External quality assurance (EQA) of screening programmes is delivered using Healthcare Improvement Scotland's quality of care approach and the Quality

Framework.[5] This approach specifies how Healthcare Improvement Scotland will design and deliver EQA activity to support improvement in healthcare.

The quality of care approach emphasises the importance of regular open and honest self-evaluation using the Quality Framework as a basis, combined with other relevant data and intelligence including the performance against these standards. Any outcomes from the quality assurance activity will be risk based and set in the context of the programme capacity for improvement. Further information on this approach is available on the Healthcare Improvement Scotland website (www.healthcareimprovementscotland.org).

Implementation

Healthcare Improvement Scotland develops and publishes national standards to support organisations and health professionals in providing a high quality pregnancy and newborn screening programme. The implementation of these standards is for local determination.

Summary of standards

Standard 1: All newborn babies are offered newborn bloodspot screening.

Standard 2: All newborn bloodspot sampling is timely and of high quality.

Standard 3: Second bloodspot sampling for cystic fibrosis and congenital hypothyroidism is undertaken within agreed timescales.

Standard 4: There is timely referral for investigation and management of all babies who have received a positive newborn bloodspot screen result.

Pregnancy and newborn screening: newborn bloodspot standards

Standard 1: Eligibility and coverage

Standard statement

All newborn babies are offered newborn bloodspot screening.

Rationale

Newborn bloodspot screening is offered to identify specific conditions as soon after birth as possible.[4, 5, 10]

The eligible population is the total number of babies born within the reporting period whose mothers were resident within the NHS board area or moved into the area within 12 months of birth.

Newborn bloodspot screening looks for the following serious but treatable conditions:[2, 3]

- phenylketonuria (PKU)
- congenital hypothyroidism (CHT)
- sickle cell disease (SCD)
- cystic fibrosis (CF)
- 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).

Babies who have been identified as having one of the above conditions need to start treatment as soon as possible to offer the best start in life.[4, 6]

Providing parents and carers with the most up-to-date and accurate information allows them an opportunity to make an informed decision about screening for their baby.[3, 4, 7, 11] Before testing, parents and carers should have a discussion with healthcare professionals, which should include the purpose, process and benefits of testing.[4]

When making decisions and informed choices, parents and carers should be fully involved and supported with their decisions, and choices should be respected by healthcare professionals.[1, 4, 9]

In the event that newborn bloodspot screening is declined, parents and carers should be given information on how to opt back in to the screening process, should they change their mind.[3, 4, 12] Information should also be provided on any signs or

symptoms which may relate to conditions that may have been picked up at screening.[3, 4, 12]

Criteria

1.1 NHS boards have systems and protocols in place to ensure all newborn babies:

- are registered within the NHS board of residence
- are recorded on the Child Health Information System (CHIS) within the NHS board of residence, and
- have processes in place with the child health/screening department to identify and follow up children with no recorded screening outcome.

1.2 NHS boards have systems and protocols in place for timely and person-centred newborn bloodspot screening, which includes:

- timescales and methods for communication of results
- providing opportunities to discuss with parents and carers the results, further management and/or further testing
- appropriate management for all resident babies who were born outwith their NHS board area, and
- processes for follow-up diagnostic testing.

1.3 Parents and carers receive information about newborn bloodspot screening at least 48 hours in advance of the screening test.

1.4 NHS boards offer timely newborn bloodspot screening to:

- all newborn babies ideally between 96–120 hours after birth, and
- all babies who have moved into the area within 12 months of birth.

1.5 NHS boards have a failsafe protocol for all resident babies to ensure that all babies on CHIS have a newborn bloodspot outcome recorded and follow-up action is taken as required.

1.6 The decision to accept or decline newborn bloodspot screening is recorded and shared appropriately.

What does the standard mean for parents and carers of the screened baby?

- Parents and carers will:
  - be offered newborn bloodspot screening at the appropriate time
  - be advised of the newborn bloodspot screening pathway
  - be offered the opportunity to discuss newborn bloodspot screening and results with an appropriate healthcare professional and all decisions will be respected, and
  - have their baby's newborn bloodspot test completed within the agreed timescales.

What does the standard mean for staff?

- Staff can demonstrate knowledge of the:
  - importance of the newborn bloodspot screening programme
  - eligibility criteria for offering newborn bloodspot screening, including movers in to the NHS board area
  - patient pathway
  - process for management of babies whose parents and carers have declined screening, and
  - benefits and limitations of screening.
- Staff will:
  - provide information and support to parents and carers that is sensitive and respects their choices, and
  - ensure decisions to accept or decline screening and/or referral for appropriate assessment are recorded and shared appropriately.

What does the standard mean for the NHS board?

- The NHS board will:
  - have an effective system in place to ensure all babies are offered bloodspot screening within the agreed timescales
  - regularly check the Child Health Information System to identify babies with a result that requires follow-up, and
  - monitor the newborn bloodspot screening pathway to ensure it is timely and completed.

Practical examples of evidence of achievement *(NOTE: this list is not exhaustive)*

- Monitoring reports detailing completion of the screening pathway.
- Protocols for eligibility and invitation to the newborn bloodspot screening service.
- Evidence that a baby undergoes screening if parents and carers wish this.
- National laboratory reports.
- Child Health Information System reporting results at the 6–8 week child health review.

Standard 2: Sampling

Standard statement

All newborn bloodspot sampling is timely and of high quality.

Rationale

The aim of this standard is to ensure that a high quality bloodspot sample is taken at the right time. Evidence shows that samples should be taken from babies 96–120 hours (4–5 days) after birth.[2-5] Babies who are premature, unwell or have had a blood transfusion should also have a sample taken; this information should be included on their blood spot card.[3, 7]

Obtaining high quality blood spot samples ensures that babies with a positive screen are identified, referred and treated early.[7, 12]

A high quality sample is one that:

- contains accurately completed data fields to enable identification of the baby, the analysis and reporting of results[7, 12]
- contains sufficient blood to perform all tests (each circle filled and evenly saturated by a single drop of blood that soaks all the way through the bloodspot card)[7, 12]
- is not contaminated,[7, 12] and
- arrives in the laboratory in a timely manner.[7, 10]

Criteria

2.1 Prior to the sample being taken, parents and carers have:

- a pre-test discussion covering the purpose, process and benefits of the test (with the healthcare professional taking the sample), and
- provided consent.

2.2 To ensure timely screening, all data fields on the screening card must be accurately completed, including the baby's community health index number.

2.3 Newborn bloodspot sampling is undertaken by appropriately trained staff within agreed national protocols.

2.4 NHS boards have protocols in place to minimise the number of avoidable repeated bloodspot samples.

2.5 NHS boards have a failsafe protocol in place for babies registered on the Child Health Information System to ensure that:

- babies with no screening result recorded by day 18 are immediately followed up to determine whether they require bloodspot testing or retesting

- if retesting is required, this is undertaken as soon as possible, and
- repeat test requests are followed up to ensure a high quality bloodspot sample is taken and a result is available.

What does the standard mean for parents and carers of the screened baby?

- Parents and carers will:
  - be offered newborn bloodspot screening for their baby at the appropriate time
  - have a high quality blood sample taken within the agreed timescales
  - be fully informed about all aspects of the screening process, and
  - be offered the opportunity to discuss newborn bloodspot screening with an appropriate healthcare professional, and
  - have their decisions respected.

What does the standard mean for staff?

- Staff can demonstrate knowledge of:
  - the patient pathway
  - how to access the Child Health Information (CHI) number for all babies undergoing screening, and
  - what a high quality sample consists of for processing.

What does the standard mean for the NHS board?

- The NHS board will:
  - ensure continued professional development relevant to staff roles is monitored and access is provided to approved training
  - have an effective system in place to ensure all babies are offered bloodspot screening within the agreed timescales
  - have a primary and failsafe mechanism in place to ensure that results are received for all screened newborn babies
  - provide parents and carers with information and support where appropriate
  - regularly check the Child Health Information System to identify babies with no screening outcome recorded and ensure that a repeat test is requested without delay, and
  - monitor the newborn bloodspot screening pathway.

Practical examples of evidence of achievement *(NOTE: this list is not exhaustive)*

- Training records and evidence of ongoing continued professional development.
- Documentation demonstrating discussions, decision-making and information is shared appropriately between teams.
- Monitoring reports detailing completion of screening pathway.
- Protocols for eligibility and invitation to the newborn bloodspot screening service.
- Evidence that a baby undergoes screening if parents and carers wish this.
- National laboratory reports.

- Child Health Information System reporting results at the 6–8 week child health review.

Standard 3: Second sampling for cystic fibrosis and congenital hypothyroidism

Standard statement

Second bloodspot sampling for cystic fibrosis and congenital hypothyroidism is undertaken within agreed timescales.

Rationale

In some circumstances, the first blood spot screening sample may fail to give a clear result for cystic fibrosis (CF) or congenital hypothyroidism (CHT), therefore a second bloodspot sample is taken for further testing. It is important that the second bloodspot sample is taken within the agreed timescales in line with national guidance.[7]

Before the second sample is collected parents and carers should be given the most up-to-date and accurate information to allow them an opportunity to make an informed decision about screening for their baby.[7, 11, 13] When making decisions and informed choices, parents and carers should be fully involved and supported, with their decisions and choices respected by healthcare professionals.[7, 11]

CF is an inherited condition that affects one in every 2,500 babies in Scotland and affects most parts of the body, especially the lungs and pancreas, and can cause regular chest infections and problems with digesting food and absorbing the nutrients needed.[4] CF gets slowly worse over time as it damages the lungs and digestive system. Children with CF are at higher risk of developing other conditions such as osteoporosis or diabetes.[14] Evidence shows that, where there is an inconclusive result, the repeat sample should be taken between day 21 and day 24 (21-28 acceptable).[2]

CHT affects one in every 3,500 babies in Scotland. Babies with CHT do not produce enough thyroxine, which is needed for mental and physical development.[4, 13] Evidence shows that where there has been a borderline thyroid-stimulating hormone (TSH) result there should be 7–10 calendar days between samples to detect changes to the TSH levels.[7, 13]

Criteria

3.1 For cystic fibrosis, a second bloodspot sample (for raised immunoreactive trypsinogen) is taken between day 21 and day 24 (21-28 acceptable), where day of birth is day 0.

3.2 For congenital hypothyroidism, a second bloodspot sample for borderline thyroid-stimulating hormone is taken between 7–10 calendar days after the initial borderline sample.

3.3 For premature babies, a second bloodspot sample for congenital hypothyroidism is undertaken on day 28, where day of birth is day 0, or on discharge from neonatal care, whichever is soonest.

3.4 Second bloodspot screening samples for cystic fibrosis and congenital hypothyroidism are taken in line with national guidance.

What does the standard mean for the individual participating in newborn screening?

- Parents and carers will:
  - have a high quality second bloodspot sample taken from their baby if required within the agreed timescales
  - be fully informed about all aspects of the screening process, and
  - be offered the opportunity to discuss a second screening with the appropriate healthcare professional, and
  - have their decisions respected.

What does the standard mean for staff?

- Staff can demonstrate knowledge of the:
  - eligibility criteria and timing for second sampling
  - patient pathway
  - process for opting in and opting out of the newborn bloodspot screening, and
  - benefits and limitations of screening.
- Staff will provide information and support to parents and carers that is sensitive to their needs and respects their choices.

What does the standard mean for the NHS board?

- The NHS board will:
  - have an effective system in place to make sure a high quality second bloodspot sample is taken from babies at the correct time period if required, and
  - ensure continued professional development relevant to staff roles is monitored and access is provided to approved training.

Practical examples of evidence of achievement *(NOTE: this list is not exhaustive)*

- Documentation demonstrating discussions, decision-making and information is shared appropriately between teams.
- Monitoring reports detailing completion of the screening pathway within an agreed and defined reporting period.
- Evidence that a baby undergoes screening if parents and carers wish this.
- Training records and evidence of ongoing continued professional development.
- National laboratory reports.

Standard 4: Referral

Standard statement

There is timely referral for investigation and management of all babies who have received a positive newborn bloodspot screen result.

Rationale

Babies who have been identified as having one of the screened for conditions should start treatment as soon as possible to offer the best start in life[5, 7] and should be referred to the appropriate service or team immediately.[2]

Where a diagnosis of any of the conditions is identified, information on the condition should be made available to parents and carers. Providing parents and carers with the most up-to-date and accurate information allows them an opportunity to make an informed decision about further treatment for their baby.[2, 7, 11] When making decisions and informed choices, parents and carers should be fully involved and supported, with their decisions and choices respected by healthcare professionals.[2, 7, 11]

Criteria

4.1 All babies with positive screening results are appropriately referred to specialist services within nationally agreed timescales.

4.2 NHS boards initiate specialist care within the specified timescales for the screened condition set by the Scottish pregnancy and newborn screening programme.[15]

What does the standard mean for the individual participating in newborn screening?

- Parents and carers will be confident that their newborn baby will be:
  - referred for investigation, treatment and management within the agreed timescales, and
  - offered the most appropriate care, and/or treatment in the event of a positive result.

What does the standard mean for staff?

- Staff can demonstrate knowledge of the:
  - specific tests and timescales, and
  - patient pathway.
- Staff will provide information and support to parents and carers that is sensitive and respects their choices.

What does the standard mean for the NHS board?
• The NHS board will: - ensure that standards of care for the delivery of screening are in place and are monitored for compliance to national standards and guidance, and - monitor the newborn bloodspot screening pathway to ensure it is timely.
Practical examples of evidence of achievement *(NOTE: this list is not exhaustive)*
• Documentation demonstrating discussions, decision-making and information is shared appropriately between teams. • Evidence that all treatment is initiated within defined timescales. • Monitoring reports detailing completion of the bloodspot screening pathway within an agreed defined reporting period. • National laboratory reports.

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