

Pregnancy and Newborn Screening:

Haemoglobinopathies in Pregnancy Standards

January 2019

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.

Haemoglobinopathies screening in pregnancy

The screening test is a simple blood test, with minimal risk to the woman or her unborn baby. At, or shortly after, the first midwife visit the woman is offered screening tests for sickle cell and thalassaemia disorders. These conditions are sometimes referred to as haemoglobinopathies. These tests should be carried out as early as possible in pregnancy – ideally by 10 weeks gestation.²

The aim of offering haemoglobinopathies screening in pregnancy is to identify the biological parents who are at risk of having an affected child and offer them information on which to base future reproductive choices.³

There are three groups of women who are at a higher chance of having an affected pregnancy:

- those where both biological parents are carriers of haemoglobinopathies
- those where the mother has a significant haemoglobin variant and the biological father's status is unknown, and
- those with donor egg or sperm where the biological parent is a carrier.⁴

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)⁵ to document progress in areas that have been identified within the pregnancy and newborn screening programme as requiring additional support.

These standards should be read alongside other relevant legislation, standards and guidance such as the KPIs mentioned above.

Scope of standards

These standards apply to screening for haemoglobinopathies in pregnancy and cover:

- eligibility and coverage, and
- screening and diagnosis.

These haemoglobinopathies in pregnancy 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 first antenatal contact can be defined as the first appointment with the midwife or hospital.

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 eligible pregnant women are offered haemoglobinopathies screening.

Standard 2: Pregnancy screening for haemoglobinopathies is safe, effective and person centred.

Pregnancy and newborn screening: haemoglobinopathies in pregnancy standards

Standard 1: Eligibility and coverage

Standard statement

All eligible pregnant women are offered haemoglobinopathies screening.

Rationale

Haemoglobinopathies are a group of inherited blood disorders that involve an abnormality in the structure of haemoglobin, a protein responsible for transporting oxygen in the blood. Haemoglobinopathies are common in people whose family origins are in parts of the world where malaria is endemic.⁴ In the UK, they can be found more commonly among people from minority ethnic groups from Africa, the Caribbean, the Mediterranean, South East Asia, the Middle East and the Far East.^{3, 7, 8} They can have a significant impact on an individual's health and wellbeing, and can restrict a person's ability to conduct normal day-to-day activities.^{3, 7}

In Scotland, all women should be offered screening for sickle cell and thalassaemia by 10+0 weeks gestation.^{1, 3, 4, 7, 8} This allows time for further screening, if necessary, and for individuals to make an informed choice about further testing and/or management.⁷

The provision of information about screening tests is essential to support women and, where appropriate, their partners to make an informed and considered decision about whether or not to undergo haemoglobinopathies screening.^{1, 3, 8} Good communication between individuals and their healthcare provider is vital as the way that screening is offered can impact the decisions made.¹ Individuals should be fully involved and supported as outlined within the general standards, with their decisions and choices respected by healthcare professionals.^{1, 3, 9}

Efforts should be made to ensure all individuals understand the purpose, benefits, limitations and implications of screening. Where an individual declines screening for haemoglobinopathies this should be recorded. Information on how to notify the screening service should they change their mind and wish to opt back in should be explained.^{3, 4}

Criteria

- 1.1** NHS boards have systems and processes in place to identify and invite all eligible women at the appropriate stage of their pregnancy for screening for haemoglobinopathies.
- 1.2** At the first antenatal contact:
 - all eligible women are offered screening for haemoglobinopathies, and
 - the decision to accept or decline screening and/or referral for appropriate assessment is recorded and shared appropriately.

- 1.3** All women receive information about haemoglobinopathies screening in pregnancy at least 48 hours before their first contact with the midwife or hospital service.
- 1.4** NHS boards have systems and protocols in place for timely and person-centred haemoglobinopathies screening in pregnancy, which includes:
- identification of eligible women
 - offering haemoglobinopathies screening at the most appropriate stage in pregnancy
 - advice and support for individuals presenting after an appropriate stage of pregnancy for screening
 - timescales and methods for communication of results
 - discussing the results with women, further management and/or further testing, and
 - a process for follow-up diagnostic testing.

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

- Women will be offered haemoglobinopathies screening at the appropriate time during their pregnancy.
- Women will receive information that:
 - is in a style and format appropriate to their needs and at least 48 hours before their first contact with the midwife
 - provides an overview of the screening process, and
 - outlines the benefits and limitations associated with screening for haemoglobinopathies.
- Women will be offered an opportunity to discuss screening for haemoglobinopathies with an appropriate healthcare professional to allow them to make an informed choice that will be respected.

What does the standard mean for staff?

- Staff can demonstrate knowledge of:
 - eligibility criteria for offering haemoglobinopathies screening to pregnant women and biological fathers (where antenatal screening shows the mother is a genetic carrier)
 - the patient pathway
 - how the individual can opt in and opt out of haemoglobinopathies screening, and
 - recording the woman's decision on haemoglobinopathies screening in electronic notes and sharing appropriately.

- Staff will:
 - provide information and support to all eligible women that is sensitive and respects their choices, and
 - ensure decisions to accept or decline screening and/or referral for appropriate assessment is recorded and shared appropriately.

What does the standard mean for the NHS board?

- The NHS board will:
 - have an effective system in place to invite eligible women for haemoglobinopathies screening at the appropriate time in their pregnancy, and
 - monitor the haemoglobinopathies screening pathway to ensure it is timely.

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

- Monitoring reports detailing completion of the haemoglobinopathies screening pathway within an agreed and defined reporting period.
- Protocols for eligibility and invitation to the haemoglobinopathies screening service.
- Protocols for minimising barriers with reference to local population.
- Evidence that any woman who wishes to have screening undergoes screening.

Standard 2: Screening and diagnosis

Standard statement

Pregnancy screening for haemoglobinopathies is safe, effective and person centred.

Rationale

Where a woman has been found to be affected or is a carrier (screen positive), it is recommended that the baby's biological father is also tested to establish their status^{1, 7, 10} and obtain a more accurate result of the baby's chances of having either sickle cell or thalassaemia.^{3, 8} Timely screening of the biological father should be offered, where appropriate, to allow for an offer of prenatal diagnosis in higher chance pregnancies before 12+0 weeks gestation. If accepted, the prenatal diagnostic test (PND) should be performed before 12+6 weeks gestation.^{4, 8, 10, 11}

If both genetic parents are carriers of a significant haemoglobinopathy, the baby has a one in four risk of being affected by a major haemoglobinopathy with a significant impact on quality of life.¹²

Individuals who have been identified as being at risk of having a baby with either sickle cell or thalassaemia should be offered counselling and diagnostic testing for the baby.^{1, 4, 8, 10} Information on the PND test should be provided to allow them an opportunity to make an informed decision about further treatment and/or whether to continue with the pregnancy.^{1, 3, 4, 10}

When making decisions and informed choices, women and, where appropriate, partners should be fully involved and supported, with their decisions and choices respected by healthcare professionals.^{1, 3, 9}

Where the baby's biological father is not able or willing to undergo testing, pregnancies should be considered as high risk.¹⁰

It is important that testing is done in a timely manner to allow fully informed choice.

Criteria

- 2.1** All women presenting before 10+0 weeks gestation are offered screening for haemoglobinopathies by 10+0 weeks gestation.
- 2.2** All women presenting after 10+0 weeks gestation are offered screening for haemoglobinopathies as soon as possible.
- 2.3** At the first antenatal contact for all women:
 - all relevant fields of a Family Origin Questionnaire (FOQ) are completed, and
 - a full blood count is taken.

- 2.4** Screening results are issued to the maternity service within 3 working days of receipt of the sample at the laboratory.
- 2.5** Where a screen positive result has been confirmed:
- the results are communicated to the woman within 3 working days of receiving the result, and
 - a face-to-face discussion with a screening midwife or other clinician is offered.
- 2.6** Once a carrier result is confirmed:
- the biological father will be offered testing as soon as possible after the mother's confirmation, and before 12+0 weeks gestation
 - the biological father will be offered testing, even if they have been tested previously, unless the results are available and performed within a UK accredited laboratory
 - prenatal diagnosis is offered by 12+0 weeks gestation, and
 - if the woman or biological father decline partner testing, the reason for declining should be recorded.
- 2.7** Where both biological parents are found to be carriers or affected, support and information is offered in a style and format appropriate to their needs, to allow an informed choice to be made for further management options, including further testing.
- 2.8** Where women are found to be carriers or affected, but the biological father has unknown status:
- the maternity record is updated to record this information, and
 - the newborn baby is tested during the newborn bloodspot screening test.
- 2.9** All women, who accept the offer, will have PND testing performed by 12+6 weeks gestation.
- 2.10** When the PND testing confirms that the baby has a haemoglobinopathy the NHS board will provide:
- timely specific information about the haemoglobinopathy diagnosed
 - information about living with and supporting an affected child
 - an opportunity to make an informed choice about continuing with the pregnancy with the appropriate healthcare professional, and
 - a timely management plan.

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

- Individuals will:
 - be offered screening for haemoglobinopathies as early as possible during pregnancy
 - be offered a PND test where appropriate within the agreed timescales
 - feel supported when making an informed choice about the screening process and pregnancy choices
 - be offered an opportunity to discuss haemoglobinopathies screening with an appropriate healthcare professional to facilitate an informed choice, and
 - have their informed choice supported and respected by healthcare professionals.

What does the standard mean for staff?

- Staff demonstrate knowledge of:
 - the eligibility criteria for offering haemoglobinopathies screening
 - the patient pathway
 - how to complete the Family Origin Questionnaire
 - what is being screened for during haemoglobinopathies screening, and
 - the importance of timeliness.
- Staff will:
 - provide information and support to all eligible women and biological fathers that is sensitive and respects their choices, and
 - ensure decisions to accept or decline screening and/or referral for appropriate assessment is recorded and shared appropriately.

What does the standard mean for the NHS board?

- The NHS board will:
 - have an effective system in place to invite eligible women and, where appropriate, biological fathers for haemoglobinopathies screening at the appropriate time in pregnancy
 - monitor the haemoglobinopathies screening pathway to ensure it is timely, and
 - have a primary and failsafe mechanism in place to ensure that results are received for all pregnant women screened.

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

- Evidence of completeness of the Family Origin Questionnaire.
- Monitoring reports detailing completion of the haemoglobinopathies screening pathway within an agreed and defined reporting period.
- Documentation demonstrating discussions, decision-making and information is shared appropriately between teams.
- Protocols for eligibility and invitation to the haemoglobinopathies screening service.

- Evidence that any individual who wishes to have haemoglobinopathies screening undergoes the screening.
- Audit of quality assurance of haemoglobinopathies screening tests.

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