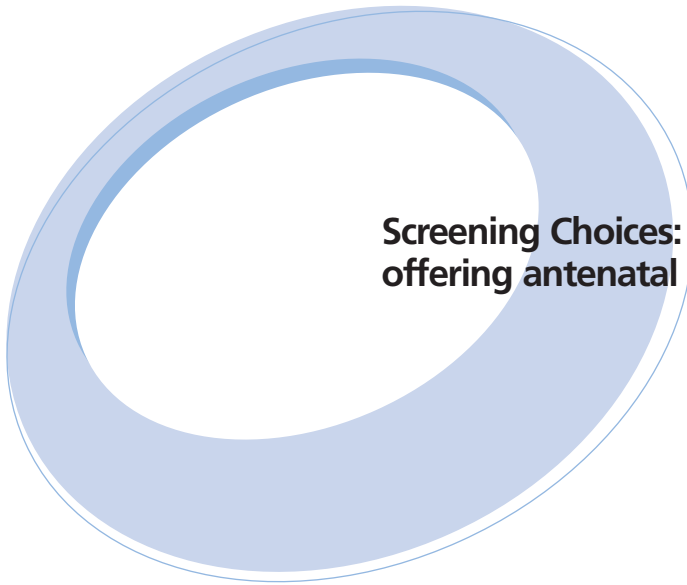


Screening Choices: A resource for health professionals offering antenatal and newborn care

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## Screening Choices: A resource for health professionals offering antenatal and newborn care

This resource has been prepared by

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On behalf of the  
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## Screening in antenatal and newborn care

# Creating a workbook



Recording your learning as you work through this unit will enable you to use it as evidence of your professional development. We have developed a workbook for this unit that repeats the activities to help you to structure your work. You can either print out the workbook if you prefer to work on paper, or save it onto your computer to work on screen.

When you have finished the unit you can keep the completed workbook in your professional portfolio with the certificate at the end of the unit, which you can sign to acknowledge your learning.

### We recommend that you create your workbook before you begin studying the unit.

To create your workbook go to Toolbox, click on **Screening in antenatal and newborn care** then click **On-screen workbook** or **Printed workbook**, depending on how you intend to use it.

## Screening in antenatal and newborn care

# Introduction

Welcome to *Screening in antenatal and newborn care*, part of the UK National Screening Committee's training programme *Screening Choices: A resource for health professionals offering antenatal and newborn care*. The programme has been developed to meet the needs of all health professionals whose role impacts on antenatal and newborn screening. It aims to help you to develop the skills and knowledge you need to enable you to ensure women, their partners and families are offered informed choices in antenatal and newborn screening. For information on how to use these materials, please read the Introduction to the whole programme.

This unit focuses on screening as it applies to antenatal and newborn care, and also discusses diagnostic tests offered to pregnant women and their babies. It aims to explore the principles of, and rationale for, screening in general, and to enable you to reflect on some of the issues associated with antenatal and newborn screening. It should take you about two hours to work through at core level.

Although this material focuses on screening much of it will apply more widely to your professional practice. It aims to explore different aspects of screening, and to enable you to reflect on some of the issues associated with antenatal and newborn screening.

### Learning outcomes

After studying this unit you will be able to:

- Discuss the purposes of screening programmes and how they can be realised
- Explain what is meant by the term screening and the concepts of 'screening' and 'diagnostic' tests
- Define the following technical terms used in screening programmes: *sensitivity, specificity, false positive, false negative, detection rate*
- Find information on the current national programmes, local programmes and arrangements, and specific screening programmes
- Understand the importance of quality assurance in screening programmes and the organisation involved in ensuring this
- Describe a screening programme in the context of antenatal and newborn care
- Explain your own role within an antenatal or newborn screening programme.

Before you begin to work through the unit, Activity 1 will enable you to evaluate your skills and knowledge in relation to screening in your practice, and to recognise the areas you may need to focus on in particular. You may find that some areas are more relevant to you than others, and decide to study those at the advanced level after working through the unit at core level.

## Activity 1



Use your workbook to do this activity

## Self-assessment

This programme has been designed so you can build on your current skills, and to provide you with a framework to structure the new knowledge and skills you develop. Our intention is that you will work through the entire unit at core level, rather than use it to 'dip into'. You can then go back to work through some or all of it at advanced level if you feel it is appropriate for your practice. Studying the units in this way will enrich your existing knowledge, and help to apply it to your practice more effectively.

As a health professional busy 'doing the job' it can be difficult to stand back and identify the skills and knowledge you use in your everyday practice. To get the most out of studying this unit it is good to be aware of the extent of your existing knowledge.

The self-assessment below will help you to think about your knowledge and skills about screening. It is important to remember that this is simply an exercise to help you plan your learning. There are no 'right' or 'wrong' answers, and the responses you give are for your eyes only.

### The self-assessment

Look at the seven statements below and in your workbook rank how much you know about each, and how competent you feel about completing the task(s) involved in them. Use a scale of 1–5 (1 being NOT very competent and 5 being VERY competent).

1. I can discuss the purposes of screening programmes and how they can be realised.
2. I can explain what is meant by the term 'screening' and the concepts of screening and diagnostic tests.
3. I can define the following technical terms used in screening programmes easily and in a way that women, their partners and others can understand:
  - Sensitivity
  - Specificity
  - False positive
  - False negative
  - Detection rate.
4. I can find information on:
  - Current national programmes
  - Local programmes and arrangements
  - Specific screening programmes.
5. I understand the importance of quality assurance in screening programmes and the organisation involved in ensuring this.
6. I can describe a screening programme in the context of antenatal and newborn care.
7. I can explain my own role within an antenatal or newborn screening programme.

### Identifying your skills and knowledge

Use the scores you gave yourself for each statement to identify the areas of your practice you feel least competent about and where you are very confident. You might like to make some brief notes about what you would like to learn from studying the unit and how it could enhance the care you provide.

The unit begins by looking at how health screening is organised in the NHS and at the nature of health screening in general. It discusses why screening programmes are developed, the limitations of screening, what elements are necessary to ensure they are effective and how they are monitored. It then goes on to look more specifically at antenatal and newborn screening and enables you to consider your own role in antenatal and newborn screening services.

Before you work through the unit, it is important to understand the standards of service all screening programmes in antenatal and newborn care must achieve (see box). These are set out in the *National Service Framework for Children, Young People and Maternity Services* (DH, 2005) and *Policies and Standards for Newborn Blood Spot Screening* (<http://www.newbornscreening-bloodspot.org.uk>). Another important point is that while screening programmes focus on investigating the risk of, or diagnosing, specific conditions in women and their babies the philosophy of maternity services is based on treating pregnancy and birth as normal events until they are proven otherwise.

**Think point:  
involving parents**

*'The ultrasonographer did a wonderful job of explaining everything and taking her time. I mean, you're looking at something that's black, white and various shades of grey, and it's not the easiest thing to make out. I was probably looking at an eye when I was supposed to be looking at a nose, things like that. We didn't feel we just happened to be there, she made us feel we were a part of it and that we had some sort of say, and that we had brains, which was nice.'*

Parent

All NHS maternity care providers and primary care trusts must ensure that:

- A comprehensive, high-quality antenatal screening and diagnostic service, based on current recommendations of the National Screening Committee (NSC) and designed to detect maternal or fetal problems at an early stage, is offered to all women
- Antenatal tests and screening are offered to women as options (with the purpose and consequence of each test explained), rather than as a routine part of the process of being pregnant
- Staff working with women in the preconception and antenatal period are competent in recognising, advising and referring women who would benefit from more specialist services
- All relevant clinical guidelines from the National Institute for Clinical Excellence are followed, such as the *Guidelines for Routine Antenatal Care*
- Where women request or decline services or treatment, their decision is respected
- All newborn infants have a clinical examination to detect preclinical abnormalities within the first week of life for full-term babies, or prior to discharge home from neonatal care and a further physical examination at six to eight weeks
- All newborn babies must be offered newborn blood spot screening, which is recommended, and this should be performed when the baby is 5-8 days old
- Professionals are skilled in sharing concerns and choices with parents as part of the emerging diagnosis
- Both parents are encouraged to be present at the first examination
- Professional staff examining newborn babies have up-to-date training in neonatal examination techniques
- Prompt referral for further medical examination or treatment is provided through agreed clinical care pathways.

## Screening programmes in the NHS

The purpose of screening is the early detection and prevention of disease. To many people, public and professionals alike, it is a very attractive idea. However, developing effective screening programmes is a complex exercise and requires a great deal of preliminary analysis, planning and careful implementation.

Screening programmes offered by the NHS are overseen by the UK National Screening Committee (NSC). This is part of the structure of the NHS and advises on:

- The case for new programmes
- The introduction and implementation of new screening programmes
- The case for continuing, modifying or changing existing screening programmes.

You can find out more about the NSC on its website (<http://www.nsc.nhs.uk>)

As you know, the UK has a number of formal screening programmes, which only came into place after much public and professional debate. In order to structure this debate, formal criteria have been developed against which potential and actual screening programmes are judged. While few conditions will meet all the criteria, the final decision about whether or not a screening programme should be developed will depend on how well a condition meets the majority of the criteria.

It is important for you to understand how these decisions are made, so that you can help women and families to understand why some screening tests are available in national programmes and, equally importantly, why others are not.

Antenatal and newborn screening has the potential to estimate risk for, or enable the detection of, far more conditions than national programmes cover. You therefore need to be able to explain why particular conditions are not universally screened for, and why some people may be offered tests that are not offered to others.

Decisions made by the NSC are policy decisions, but as a practitioner you need to understand the criteria the NSC uses in trying to reach decisions. They relate to:

- The condition to be screened for
- The screening test
- The treatment available
- The screening programme.

Activity 2 will help you to understand how the criteria can be used to make decisions about programmes.

## Activity 2

### Criteria for screening programmes

Look at the Factsheet at the end of this unit and at the criteria used by the NSC to judge whether a screening programme can be justified (<http://www.nsc.nhs.uk/pdfs/criteria.pdf>), and the discussion of the criteria on the Factsheet.

#### Core

Choose a screening test that is relevant to your work and check it against the criteria used to select national screening programmes. Make a note of the main reasons you think it was selected.

#### Advanced

Think of a condition for which a test is available but is currently not offered to all pregnant women or newborn babies. Go through the NSC criteria again and see how many the condition and test meet. You may need to research the condition further to get all the information you need.



Use your workbook to do this activity

---

## Feedback

### Core

Hopefully, you could see that the screening programme you chose to examine meets most of the NSC criteria. For example, if you looked at the newborn hearing screening programme you might have said:

- Hearing problems can cause significant problems that impact on many areas of a child's life including communication, development and personal safety, and can also affect the child's family
- If hearing impairment is detected in newborns, early intervention can have benefits in speech, social and emotional development
- Inborn hearing impairment cannot be prevented
- There are safe and effective tests to detect signs of possible hearing impairment in newborn babies
- There are well-established interventions to help children with hearing impairment, which can be started from an early age.

Further information on the newborn hearing screening programme is available on the programme website (<http://www.nhsp.info>)

### Advanced

There are numerous conditions for which tests are available but for which there is currently no national screening programme – although certain mothers or babies may be offered the tests if they are known to be at risk of the condition. When checking your chosen condition against the NSC criteria you may have found that it met many of them. However, it is likely that the criteria that were not met were significant. For example, the condition may be too rare to justify a national screening programme, or there may not be an effective intervention that can cure or reduce the effects of the condition. Could you explain to a parent why a programme is not available if you were asked?

---

Having considered screening programmes in the NHS and the role of the NSC, the next section looks at screening itself. It aims to help you to understand the nature of health screening and why you need a good understanding of the screening programmes offered by your service.

## What is screening?

People usually think of screening as finding a disease early, or even finding early warning signs before the disease appears. This has the potential to provide important and long-lasting benefits, as the two examples below illustrate:

- The early diagnosis and treatment of phenylketonuria (PKU) has reduced the risk of neurological impairment in babies born with the condition from about 80–90% to 6–8% of cases (Health Technology Assessment, 1997)
- Incidence of cervical cancer in England and Wales fell by 42% between 1988 and 1997. This fall is directly related to the cervical cancer screening programme (NHS Cervical Screening Programme, 2004). Cervical screening is thought to prevent around 1,300 deaths and up to 3,900 cases of cervical cancer per year in the UK (Sasieni, 1996; Sasieni and Adams, 1999).

Although the examples above demonstrate the potential benefits of screening, when people decide whether to accept or decline it they need to understand the reasons for the test and the meaning of the result – both in terms of its reliability and its implications for them – in order to make an informed choice. If they do not fully understand these issues they are unable to make an informed choice, as the quote by Kate illustrates. To facilitate this, you need to be able to offer them the information, advice and support they need.

*'Women often come along for the 20-week scan with no idea what it's for other than to get a picture of their baby and to find out if it's a boy or a girl. They don't understand that it's a screening test in itself. Some have declined all the other tests but they'll have this one because they'll get the picture.'*  
Kate, ultrasonographer

In order to enable women, their partners and families to make informed choices about offers of antenatal and newborn screening you need to understand the concept of screening in general. This is quite complex – look at the NSC’s definition below:

*Screening is a public health service in which members of a defined population, who do not necessarily perceive they are at risk of, or are already affected by a disease or its complications, are asked a question or offered a test, to identify those individuals who are more likely to be helped than harmed by further tests or treatment to reduce the risk of a disease or its complications.*

(UK National Screening Committee: <http://www.nsc.nhs.uk>)

If you examine the NSC’s definition carefully you will see that screening has a number of important elements. The key ones are:

- Screening is undergone by ‘well’ people who might not even have thought about the disorder or problem in question
- Screening is a systematic activity – it is a programme, not just the offer of a single test
- Screening tests do not usually provide a final diagnosis – in most cases people whose results suggest they are at an increased risk of developing the condition in question are offered diagnostic tests or treatments to reduce their risk
- A screening programme is applied to a whole group – systems are set up to ensure all those in the target group are offered screening – they do not have to know about the programme and ask for the test themselves.

Now look at how two screening programmes work to prevent disease or detect it early in order to minimise the harm it causes.

### **Cervical screening – detects precancerous cells**

Cervical screening is not a test for cancer. It is a method of reducing risk of cancer by finding and treating early abnormalities that, if left untreated, could lead to cancer in a woman’s cervix. The first stage in cervical screening is to obtain a sample of cervical cells by means of the conventional Pap smear test or the newer technique of liquid-based cytology (LBC). If precancerous cells are detected, women can be treated to remove these.

### **Phenylketonuria screening – picks up a metabolic condition before it has the chance to do harm**

Phenylketonuria (PKU) is a rare inborn error of metabolism where a baby is deficient in the enzyme phenylalanine hydroxylase. This means the baby cannot break down phenylalanine, a substance found in many high-protein foods, so it accumulates in the body. If untreated, PKU results in severe learning disabilities and neurological problems. However, timely intervention can prevent these problems.

All babies should be offered screening for PKU as part of the newborn blood spot screening programme.

The aim of screening for PKU is to detect infants with the condition early so that they can be treated before irreversible damage is done. Babies with PKU must be started on a low phenylalanine diet before they are 21 days old to ensure normal intellectual development. Current guidelines recommend that they should continue to have a low phenylalanine diet for the rest of their lives.

More information about the newborn blood spot screening programme is available on the programme's website (<http://www.newbornscreening-bloodspot.org.uk>). You can also find information on PKU on the website of the National Society for Phenylketonuria (<http://www.nspku.org>).

Some of the conditions pregnant women and newborn babies can be screened for are genetic. For example, PKU, cystic fibrosis (CF) and sickle cell disorder all fall into this category. These are autosomal recessive conditions, so if they are detected in newborn blood spot screening, both parents are carriers. This is important information for parents as it can inform future reproductive decisions.

*There is more on genetics and genetic inheritance in the unit **Understanding genetics**.*

National antenatal and newborn screening programmes have been developed with the aim of detecting a range of conditions – or risk of them – in pregnant women and their babies. The tests are offered during a woman's pregnancy, or shortly after her baby is born. Some give an indication of the risk of a specific condition, while others can diagnose with certainty whether the mother or baby is affected.

## **Terminology**

A number of terms are commonly used in relation to screening. It is important to understand these so that you can explain them to parents and families:

**Sensitivity/detection rate:** the proportion of affected people who are identified by a screening test.

**Specificity:** the proportion of unaffected people who are correctly identified by a screening test.

**True positive:** a test result that correctly shows the person is affected by or at risk of the condition in question.

**True negative:** a test result that correctly shows the person is not affected by or at risk of the condition in question.

**False positive:** a test result that shows the person is affected by or at risk of the condition in question when this is not the case.

**False negative:** a test result that shows the person is not affected by or at risk of the condition in question when this is not the case.

**Positive predictive value (PPV):** The probability that a patient has the disease/condition when given a positive result.

*There is more on the concept of risk in relation to antenatal and newborn screening in the unit **Understanding and communicating risk.***

**Think point: the opportunity to plan ahead**

*'One of the points that the doctor made that I thought was very relevant and was a good reason for amniocentesis – is the sooner you find out if you're going to have a baby that's got a problem, you can start getting your resources and your help and your support together, rather than waiting until the baby's born and having a shock. You can go through a great deal of preparation, the grieving process. All of that can happen earlier on rather than later. I think that's sensible.'*

Pregnant woman

## Diagnostic tests

Many screening programmes offer follow-up tests to those who are identified by screening to be at an increased risk of the condition in question. It is important to understand the difference between screening tests, which estimate risk, and diagnostic tests, which can confirm the absence or presence of the condition.

In the Down's syndrome screening programme, for example, the mother is first offered a dating scan and blood test that, together with her age, are used to calculate an estimated risk of her baby being affected with Down's syndrome. This is sometimes combined with other screening procedures such as nuchal translucency (NT) scanning in order to generate a 'combined' risk assessment. The Factsheet gives more information about the tests that can be offered. Women in higher-risk categories are then offered a diagnostic test that involves examination of all the baby's chromosomes (karyotype) to look for the extra chromosome 21 that is present in Down's syndrome.

*There is more on how genetics relates to antenatal and newborn screening in the units **Understanding genetics** and **Understanding and communicating risk.***

**Think point: clear information**

*'One thing to add about CVS [chorionic villus sampling] – which is not to deter people from doing it, but because we think you should have all the facts. But it was portrayed to some extent as being just a little prick in the stomach, and it was far from that. I think people should be aware that it is not just a little discomfort. I mean people have different pain thresholds, but I found it extremely painful.'*

Pregnant woman

Activity 3 will help you to understand the relationships between screening and diagnostic tests. The advanced activity examines how screening terminology applies to specific results. You need to understand these to ensure you can effectively communicate with women considering screening to ensure they are aware that the results are not absolute, and to help them understand what the test results mean for them.

### Activity 3

## Down's syndrome tests

Core

Think about women whose test results puts them in each of boxes a, b, c, and d. Describe in words what has happened and what their results mean.



Use your workbook to do this activity

		Diagnostic test (the 'truth')	
		Positive	Negative
Screening test (whether the screening test thinks you are at 'higher' or 'lower' risk)	Positive	A	B
	Negative	C	D

Advanced

Look at the figures in the example below. Try to put the right numbers into all the boxes on the grid (this is repeated in your workbook) and answer the questions below the grid.

*Example*

15,000 women have a serum screening test for Down's syndrome. 500 have screen-positive results. Diagnostic testing demonstrates 10 to have Down's syndrome. A further five babies from the initial 15,000 are delivered with Down's syndrome.

		Down's syndrome found after antenatal tests or after birth		Totals
		Positive	Negative	
Result of serum screening test (whether the screening test suggests the pregnancy is 'higher' or 'lower' risk)	Positive			
	Negative			
Totals				

### Questions

1. How many women had a baby with Down's syndrome?
2. How many of these were picked up by the serum screening test?
3. How many women had a baby that did not have Down's syndrome?
4. How many of these were rightly identified as negative by the serum screening test?

3 continued

3 continued

5. If a woman has a positive serum screening result, what is the probability of the baby having Down's syndrome?

Now look at how often the screening test gets it 'wrong':

6. How many women had a positive screening test, putting them into a high-risk group, but then were found not to have a baby with Down's syndrome?
7. How many women had a negative screening test, putting them into a low-risk group, but then gave birth to a baby with Down's syndrome?

## Feedback

Core

Your descriptions of the results for each of the four boxes should have been similar to the following:

- a. A screening test has shown that the woman's baby has a high risk of Down's syndrome, and this has been confirmed by a diagnostic test.
- b. A screening test has shown that the woman's baby has a high risk of Down's syndrome, but a diagnostic test has shown that the baby is unaffected.
- c. A screening test has shown that the woman's baby has a low risk of Down's syndrome, but a diagnostic test has found that the baby is affected.
- d. A screening test has shown that the woman's baby has a low risk of Down's syndrome, and this has been confirmed by a diagnostic test.

Advanced

		Down's syndrome found after antenatal tests or after birth		Totals
		Positive	Negative	
Result of serum screening test (whether the screening test suggests the pregnancy is 'higher' or 'lower' risk)	Positive	10	490	500
	Negative	5	14,495	14,500
Totals		15	14,985	15,000

### Answers

1. Fifteen women had a baby with Down's syndrome.
2. Ten of these were picked up by the serum screening test.  
10 out of the 15 babies with Down's syndrome were picked up as high risk by the screening test. The proportion  $10/15$  (66.7%) is known as the detection rate or sensitivity of the test.
3. 14,985 women had a baby that did not have Down's syndrome.

4. 14,495 of the 14,985 babies who did not have Down's syndrome were correctly identified as low risk by the screening test. The proportion  $14,495/14,985$  (96.7%) is known as the *specificity* of the test.
  5. If a woman has a positive serum screening result, the probability of the baby having Down's syndrome is  $10/500$  or  $1/50$ .
  6. 490 women had a positive screening test, putting them into a higher-risk group, but then were found not to have a baby with Down's syndrome.  
These cases are *false positives*. In quality assurance reports this is expressed as a *false positive rate* and is  $490/14,985$ .
  7. Five women had a negative screening test, putting them into a lower-risk group, but then gave birth to a baby with Down's syndrome.  
These cases are *false negatives*. In quality assurance reports this is expressed as a *false negative rate* and is  $5/14,500$ .
- 

Activity 3 demonstrates that screening is not an exact science. This means it also has the potential to do harm. The next section looks at the limitations of screening and the potential to do harm.

## The limitations of screening

While screening programmes are devised to benefit the population as a whole, there is a potential for individuals to be harmed. This might be caused by screening programmes themselves, as discussed below, or by people's expectations of screening as the case example on the limitations of screening illustrates. Take a look at what the NSC says about this:

*'Screening has important ethical differences from most aspects of health care, as the health service is targeting apparently healthy people, offering to help individuals to make better informed choices about their health. However, there are risks involved and it is important that people have realistic expectations of what a screening programme can deliver.'*

*'Whilst screening has the potential to save lives or improve quality of life through early diagnosis of serious conditions, it is not a foolproof process. Screening can estimate the risk of developing a condition or its complications but it cannot guarantee protection. All screening programmes have a small number of false positive results (wrongly reported as having the condition) and false negative results (wrongly reported as not having the condition) – this cannot be avoided.'* (<http://www.nsc.nhs.uk>)

While further diagnostic tests can in many cases offer a definite answer, these may be invasive and therefore have an associated risk. For example, amniocentesis and chorionic villus sampling in antenatal care are associated with a small risk of miscarriage. In addition, treatments offered when a condition is diagnosed may cause harm. For example, treatment of developmental dysplasia of the hip, which can be detected in the physical examination of newborn babies, can in a small number of cases, lead to necrosis of the hip.

The NSC is keen to ensure that people understand that screening aims to reduce risk, but that it cannot completely eliminate it.

Look again at the key elements of screening listed earlier, and how these might lead to harm for individuals who are offered screening.

- **Screening is undergone by ‘well’ people who might not even have thought about the disorder or problem in question**

*Why might this cause harm?*

By offering screening we may be creating problems in people who would never have been affected by the condition in question – for example, the test may lead to invasive investigations and, with false positive results, unnecessary treatments.

- **Screening tests do not usually provide a final diagnosis**

*Why might this cause harm?*

Some people will be misclassified due to false positive or false negative results; people may still experience anxiety after receiving their test results, or may be falsely reassured.

- **Screening is a systematic activity**

*Why might this cause harm?*

By screening the whole population, there is a risk of causing harm due to false positives or psychological harm to people by raising concerns about the possibility of the disorder in question, which, for many, will prove unnecessary.

- **A screening programme is applied to a whole group**

*Why might this cause harm?*

Some people in the target group may not be identified and offered screening.

### **Think point:**

#### **limited information**

*‘After finding our baby had Down’s syndrome we were treated with kindness and respect, and when we expressed our dread of being old and unable to cope with an adult who may have intellectual and physical impairments and may have to go into a home the consultant said: “You have obviously thought this through – most people worry about how they will cope in the early years”. We decided to end the pregnancy.’*

*‘The trouble with screening is that you only have so much information. We wanted to find out if the baby had any detectable health issues or the likely scale of the learning disability – would he suffer health problems throughout his life and not be able to understand why he was always in hospital? Of course all those questions could not be answered, and that is the problem with screening. It does not seem that there are enough answers and time is so short.’*

Parent

The next section looks at the different elements screening programmes must include to ensure they are effective and to minimise the potential to cause harm to individuals.

## The screening programme as a whole

As you have already seen, screening is more than just the administration of a simple test – it is a whole programme. For example, the newborn blood spot screening programme needs to encompass not only the offer of the test and the test itself, but also:

- The clinical services that will examine babies who are shown to have or be at increased risk of one of the conditions covered by the programme
- Any further diagnostic tests
- Interventions to treat or manage the condition in question
- Follow-up support such as counselling or health education.

Screening programmes must be able to embrace all the above aspects, as it would be unacceptable to identify babies with serious conditions but fail to provide necessary follow-up interventions.

There are many elements to a programme, which are linked to the definition of a screening programme. Look back at the definition on page 8 and notice that each programme must:

- Define the population it will be offered to – in some cases this might be people defined by age, ethnic origin or having a condition or disability who are at increased risk; we must be fair and thorough in ensuring that all members of these groups are offered and can access screening.  
(Example: all women aged between 50 and 70 years are offered screening for breast cancer.)
- Work out how each individual in the target population will be identified.  
(Example: physical examination to detect a range of conditions is offered to all babies in the first weeks of life, while they are still in the care of maternity services.)
- Work out how the offer of a test will be made.  
(Example: antenatal screening is offered to pregnant women when they attend an antenatal appointment.)
- Ensure that the result of the screening test is communicated effectively to all women, whether or not further intervention is recommended.  
(Example: results of cervical screening are sent directly to women by post.)
- Ensure fail-safe follow-up of positive tests including arrangements for subsequent diagnostic testing.  
(Example: women with a higher risk of having a baby with Down's syndrome, sickle cell disorder or thalassaemia can be offered amniocentesis, CVS or cordiocentesis.)
- Ensure that people with the disease or disorder have the necessary follow-up treatment or surveillance.  
(Example: pregnant women who test positive for HIV can be offered antiretroviral drug therapy for their own disease and advice about the safest method of delivery and feeding to reduce the risk of mother-to-baby transmission.)

As you can see, screening programmes are extremely complex to both design and operate. The next section discusses the importance of being able to ensure they are of a sufficiently high quality, and looks at the systems put in place to do this.

You can find more information about screening for sickle cell disease and thalassaemia on the NHS Sickle Cell and Thalassaemia Screening Programme website:  
<http://www.kcl-phs.org.uk/haemscreening>

### Think point: professional support

*'At my 12-week scan it was found that my baby had excess fluid in the nuchal fold. We decided to have a termination, and tests revealed the baby had Edwards' syndrome. A specialist midwife supported me and my husband through this and my subsequent pregnancy.'*

Pregnant woman

## Quality assurance

Quality assurance (QA) relates to the ways services ensure that screening programmes are of the highest possible quality and that local screening services adhere to national standards. For example, from a technical perspective professionals undertaking laboratory tests must be meticulous about issues such as how the samples are handled, machines are calibrated and results are interpreted. All the other aspects of the programme must pay equal attention to quality, including the administrative systems that ensure results are communicated to the recipients of screening. Standards documents for individual screening programmes should be available on their websites, although some of the newer programmes are still in the process of developing these. The NSC requires all programmes to have a QA process that is separate from the actual service. For example, QA in cancer screening is undertaken by teams at regional level. The QA team has the authority to regularly review and assess the process in place and make recommendations to providers, and to ensure improvements are implemented when problems are identified.

In order to measure the quality of a service, we need standards for comparison. These standards cover many aspects of the service, such as coverage (the proportion of the target population who have screening), education and training, clinical arrangements, quality of written information for patients, and technical aspects such as pathology or radiology standards. When a new programme is introduced the standards are based on background research studies, but when data become available from the programme itself the standards are reviewed and altered to reflect the realities in everyday practice. The standards of performance achieved by different services operating a screening programme will vary. These results can be used to set standards to define what is excellent performance, an achievable standard, or low level, below which no programme should fall.

QA in screening aims to help clinicians, service and programme managers reach higher standards of care by:

- Reducing the risk of errors
- Identifying, managing and learning from errors effectively and sensitively
- Helping professionals and organisations to continually improve their performance
- Setting and re-setting standards.

Activity 4 aims to help you to understand the importance of having systems to ensure the quality of screening programmes from the patient's perspective.

*There is more on how offers of screening impact on parents and families in the unit **The parent perspective on screening**.*

## Activity 4

### A high-quality programme

#### Core

Think about a screening test that you have either had or been offered, or one that you may be offered at some time – this may be from an antenatal or newborn screening programme, or another NHS programme.

Look at the following factors, and rate them from 1–5 in terms of importance to you as a possible recipient of the test, where 1 is not important and 5 is very important:



Use your workbook to do this activity

- The cost of the test
- The accuracy of the test
- Whether the test is painful
- Where the test is performed
- How quickly you receive results
- The method by which results are communicated
- Whether you can book an appointment for the test.

What other aspects of the programme would be very important to you?

#### Advanced

Look at the Department of Health's standards to support antenatal screening programme for infectious disease:

<http://www.dh.gov.uk/assetRoot/04/06/61/91/04066191.pdf>

and answer the following questions:

- Who needs to know about the standards?
- Who is the programme aimed at?
- What data are collected?
- What happens in the clinic about consent, information for women and recording of information given about the screening offered?
- Give three standards for the laboratory in rubella screening.
- How and when are results communicated to the woman – what happens to those who have a positive HIV result?

See if you can find the following:

- Who is responsible for QA in the screening programme in which you are involved?
- Is there a local or national report you could look at for the programme?
- What are the important standards, are they achieved, and if not, what is being done about it?

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## Feedback

### Core

Different people will find different aspects of a screening programme important. Some might be important to the majority of people, for example the accuracy of the tests. Other aspects, however, may be more influenced by personal experiences. For example, those who find it difficult to get time off work may think the ability to choose an appointment is important, while other people may be happy to be allocated an appointment. Discuss with colleagues what aspects are important to them – how different are other people's views to your own? How well does your service measure up against the aspects you all listed?

### Advanced

Answering the questions in the activity should have helped you to understand the complexity of the systems set up to ensure screening programmes are of high quality, and how your own service operates within the standards for the programme you are involved in. Focusing on standards may have highlighted ways your own practice or your service in general could be improved. If so, try to come up with a plan to address this. Further units in this programme or learning materials produced for the specific training programmes may help you meet any personal learning needs you identified. You may want to discuss your suggestions with your manager.

### Think point: giving choices

*'We lost our first baby due to fetal hydrops and Down's syndrome. I saw two consultants and was given the choice of a surgical or medical termination. This choice made all the difference to me. My GP had already called by the time I got home after my first scan, which was very thoughtful and much appreciated.'*

Formerly pregnant woman

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## Monitoring

Systems of QA require information about how screening programmes are operating in practice. The NSC recommends that all aspects of screening programmes are monitored and audited. This should be managed at strategic health authority or regional level.

Services providing breast and cervical screening must submit information on aspects of their performance so that their performance can be monitored. However, this requires systems to be established so that the information can be submitted easily in a consistent format. In maternity care, these systems are not well established in many parts of the country or within some screening programmes, and extracting valid and reliable data can be difficult. These systems will need to be improved over the next few years to ensure that the data required to monitor the uptake and performance of screening tests can easily be submitted and used.

So far you have considered health screening and national screening programmes in general. The unit now goes on to look more specifically at antenatal and newborn screening, and your role within screening services.

## Antenatal and newborn screening

There are important differences between antenatal and newborn screening, as you will see below.

### Antenatal screening

Antenatal screening tests can have one of three purposes:

1. To identify whether the woman has any of a range of conditions that may harm her baby unless she receives treatment during her pregnancy or the baby receives treatment shortly after birth.
2. To identify whether the baby is developing normally. It may be possible to identify conditions that may require treatment soon after birth or that may make it unlikely that the baby will survive.
3. To identify whether her baby has, or is at risk of, conditions such as neural tube defects, sickle cell disorders and thalassaemia.

**Think point:**  
**decision-making**

*'After we'd talked and talked about it I just felt, that whatever happened we'd have a baby. And it might be a baby with Down's syndrome, but that's OK. It'll still be our baby – we'll have a baby.'*

Pregnant woman

In some cases, finding out that a fetus has a particular condition allows therapy to be offered antenatally to correct or treat the condition. It can also enable maternity services to ensure the baby is delivered in the appropriate location and will have access to specialist care or treatment.

In some cases, however, there is no possible treatment, and antenatal screening identifies an increased risk of a serious disorder in the fetus. This allows prospective parents the choice of going on to have an invasive diagnostic test, after which they can decide whether to continue with the pregnancy. While some may opt for termination of pregnancy if their baby is diagnosed with a serious condition, others will continue with the pregnancy – this is the parents' choice. However, whatever they decide, it is important that they have all the relevant information and understand the implications for them, to enable them to make an informed choice.

**Think point: screening**

*'I have a fairly open mind about screening. There's always that concern that you're not getting enough information. I think there's always that feeling of "Have I missed something? Do I know the right questions to ask?" Because that's the key, isn't it? You've got to know the questions to ask, and how do you know what those questions are if you don't know the information? Or where you go to look?'*

Pregnant woman

Activities 5 and 6 will help you to reflect on your role in antenatal screening, and to look at local services in relation to the Down's syndrome screening programme. The advanced activities encourage you to reflect on the implications of antenatal screening and your own attitudes. If your role relates to newborn rather than antenatal screening, move to Activity 7.

**Activity 5**

**Your role in antenatal screening**

**Core**

Look at the list of antenatal screening tests recommended in the guidelines issued by the National Institute for Clinical Excellence (2003) on the Factsheet. Which, if any, are relevant to your work? Write down how you see your role in these programmes. Do you think your role could develop further in relation to antenatal screening? If so, what skills would you need to develop?

**Advanced**

Thinking about the screening and diagnostic tests you perform and/or discuss with women, write down the possible consequences for the mother, baby and family of receiving results that indicate a problem or increased risk of a problem of the tests you listed. Consider:

- What decisions or actions might the mother and/or family need to take?
- What social, cultural, religious or practical issues might affect their decision or action?
- How might their decision or action affect them in the future?

You may find it helpful to discuss these with a colleague to ensure you identify as many as possible. Do you feel you have the skills to help women and families to consider these consequences?



Use your workbook to do this activity

## Feedback

### Core

If you feel you need to develop further skills in order to fulfil your role in antenatal screening, you may want to work through further units in this programme, or the learning materials developed for specific screening programmes. Discuss this with your manager.

### Advanced

Having considered the possible consequences of tests for mothers, babies and families, you may feel you need to develop your skills further to help you to discuss these with the women you work with and their partners and families. You may want to work through further units in this programme such as *The parent perspective on screening* or *Informed choice for everyone: valuing diversity*, or the learning materials developed for specific screening programmes. It may be helpful to discuss this with your manager.

As you saw in the section on What is screening?, pregnant women are offered one or more screening tests which are used to give an estimate of their baby's risk of having Down's syndrome. Depending on their results, women may then be offered an invasive diagnostic test to tell them whether or not their baby is affected. Not all women are offered diagnostic tests because they are invasive and carry a small risk of miscarriage. The tests offered in different services vary, and it is important for you to know what your own service offers – Activity 6 will ensure you are clear about this.

More information on Down's syndrome screening is available on the programme website <http://www.nelh.nhs.uk/screening/dssplhome.htm> and in *National Down's Screening Programme for England: A Handbook for Staff*.

## Activity 6

### Down's syndrome screening

#### Core

Make a note of which Down's syndrome screening and diagnostic tests are available to women accessing your maternity service. Are they all part of the routine NHS package? Are there any restrictions on who is offered particular tests? What happens if a woman wants alternative tests? (If you are not sure you may need to talk to your manager about these issues.) Does your service meet the NSC/NICE guidelines listed in the Factsheet?

#### Advanced

1. Karen's Down's syndrome screening tests have estimated that she has a 1:50 risk of having a baby with Down's syndrome. What would you be able to offer her in your service, and how would you explain it to her?
2. Now imagine that Karen feels this is an acceptable risk for her, but her partner Paul does not, and wants her to have a diagnostic test. The associated risk of miscarriage means Karen does not want to do this.
  - How would you deal with this situation?
  - What if you strongly agreed with either Karen or Paul – how would you prevent your views from affecting the way you handled the situation?
  - What would you do if they asked you for your advice?



Use your workbook to do this activity

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## Feedback

### Core

Even if you are not directly involved in offering screening and diagnostic tests for Down's syndrome, you should be confident that you know all the tests offered by your service, what they involve and when they are offered, as you may be asked to explain them. If your service restricts the offer of diagnostic tests to particular groups, it is also important to know what is available for women and families who wish to pay for additional tests. If your service does not meet the NSC/NICE guidelines, you may want to discuss with your manager what developments are in place or planned, as you may be able to make suggestions.

### Advanced

Situations where there is conflict between partners about decisions related to antenatal screening and diagnostic tests can be difficult to deal with. If they ask for your help, try to tease out which specific issues are causing the conflict and encourage them to discuss these together. It can be easy to allow your personal views to affect the way you discuss tests, as the case example below demonstrates, but this is inappropriate and can compromise the ability of women and families to make informed choices.

If people ask you for advice, it is important to give this based on your professional, rather than personal opinion. You must also make it clear that this is a professional opinion based on the clinical and other information you have about their situation, and that it may not take account of other factors that are important to them. They must be clear that you can only advise, but ultimately, they must make the decision. Situations such as this can be extremely complex, and if they cannot agree on the way forward, Karen and Paul may need help from someone with counselling skills to work through their dilemma. If you do not have such skills, you should refer them to an appropriately qualified practitioner.

*You may find **The parent perspective on screening or Informed choice for everyone: valuing diversity helpful in understanding parents' situations. Getting the best out of the consultation** may help you to avoid influencing parents with your own opinions.*

*The specialist training for the Down's syndrome screening programme will help you to develop your skills and knowledge in relation to Down's syndrome*

*<http://www.nelh.nhs.uk/screening/dssp/home.htm>*

*You can also find information on the condition on the website of the Down's Syndrome Association <http://www.downs-syndrome.org.uk>*

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### Case example. Personal opinions

When Trina came for her booking-in appointment, she was already 16 weeks pregnant. She was thin and pale and her hands shook. She wasn't very forthcoming about why she presented so late, but while I was taking her history she told me she had been an injecting heroin user, but had stopped when she found that she was pregnant. I discussed this further with her emphasising the importance of her being honest for the sake of the baby. She admitted that she was still taking heroin, but said she had cut down. I arranged for her to be referred to the specialist service for women who use drugs, and continued with the booking-in.

When it came to discussing blood tests Trina declined the HIV test. She insisted that she was 'clear', and had never shared injecting equipment. I tried to explain the risks to her, and how she could be treated to reduce the risks both to herself and the baby if she was found to be positive, but she was adamant that she wouldn't have it. I must say that I found it very difficult not to put pressure on her – in fact I'm not sure that I didn't. Hopefully the specialist service will have more luck in persuading Trina. I do wonder whether my attitude came across and she reacted against it.

Sue (midwife)

## Newborn screening

By identifying conditions soon after birth, newborn screening ensures that babies affected by certain conditions can receive appropriate treatment or be referred to specialist services early. This can reduce the risk of their health being harmed, or enable service providers to organise specialist care to minimise the condition's impact on the baby's life.

Newborn screening does not present parents with the stark dilemmas they may be faced with as a result of antenatal screening. The baby has been delivered, so they do not face the possible decision of whether to terminate a pregnancy. In addition, the newborn screening programmes test for conditions that can be treated or managed, rather than to inform parents that their baby is at risk of a serious and untreatable condition.

However, while newborn screening may not seem to pose the same difficulties as antenatal screening, it can still be a source of anxiety to parents. In addition, as with all screening programmes, there is the possibility that their baby's screening test may not give the correct result. In addition, screening for CF and SCD unavoidably identifies carriers.

It is therefore just as important that parents and families have all the information and support they need to enable them to make informed choices about offers of newborn screening. If their baby is found to have one of the conditions tested for they will also need support as they take in this news and to help them understand the condition and its treatment or management.

Activity 7 should help you to appreciate some of the potential difficulties associated with newborn screening. The advanced activity encourages you to reflect on the implications of neonatal screening and how they differ from those of antenatal screening.

### Activity 7

#### Newborn screening

##### Core

Siobhan's first baby, Callum, was discharged home shortly after birth, but four days later he was admitted to the local neonatal unit with jaundice. He was discharged again after a week, having recovered, and Jaida, the midwife visited to discharge Siobhan and Callum to the care of the health visitor. While she was there Jaida asked when Callum's blood spot (formerly known as the Guthrie test) had been taken. Siobhan said he had had lots of tests and she was not sure about the blood spot. She asked what they were for, and Jaida explained the four conditions screened for and that treatment or management interventions were available for each.

When Jaida contacted the neonatal unit she found that Callum had not been tested while he was in there. Siobhan did not remember being told when she was pregnant that she would be offered blood spot tests for her baby. She was upset that Callum had missed having them taken, and worried that his health may have been compromised. The blood spot was taken immediately, and Callum was found to be healthy.

- Can you identify the factors that resulted in Siobhan not being offered blood spot screening for Callum?

##### Advanced

How do you think the implications of newborn screening differ from those of antenatal screening? How might these differences affect the way women and families make decisions about whether or not to accept screening?



Use your workbook to do this activity

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## Feedback

### Core

Two major factors are likely to have contributed to Callum missing out on the blood spot test at the usual time. Most babies are screened between five and eight days after birth. However, Callum had been discharged home then readmitted to the hospital when he was four days old. It may be that both the community service and the neonatal unit thought the other would undertake the test.

Siobhan could not remember having been told about the blood spot screening programme – if she had known about it and when to expect it she would probably have raised it with a health professional. Siobhan may have been told about the programme while she was pregnant, but as this was her first pregnancy she may have been worried about the birth or distracted by other concerns at the time. It is important that health professionals ensure parents understand what they are told about screening programmes. It is also good practice at subsequent consultations to check whether they have already been told about screening opportunities and that they understand what they are for and when they are offered.

### Advanced

In thinking about the differences between the implications of antenatal and newborn screening, you may have concluded that the implications of newborn screening relate only to the baby's health, while antenatal screening can also have implications in relation to whether or not the pregnancy is continued. In most cases decisions about whether to accept newborn screening are likely to be easier for parents, as the conditions screened for can be treated once they are identified. You may wish to discuss with colleagues how your service reacts when women or families refuse to have their babies tested.

Another important aspect of newborn screening is that blood spots are normally stored for at least five years. This contributes to the screening programme's QA and audit system and enables laboratories to double-check blood spots if a child becomes ill. Under the guidance of ethical and legal committees the blood spots can also be used for public health monitoring.

Before taking the blood spot you must make sure that parents are aware of storage and use of blood spots and alert them to the small possibility of receiving an invitation to participate in research in the future. If the parents do not wish to receive future invitations about participation in research connected to the blood spot programme, the health professional collecting the blood sample should indicate 'No research contact' on the blood spot card.

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Ensuring parents and families are able to make informed choices about offers of antenatal and newborn screening requires you to take account of their individual needs and to understand what factors may affect their needs for information, support and advice. The next section looks at how individual factors can affect parents' needs.

## Individual needs

### Think point: providing support

*'The consultant obstetrician and midwife at the fetal care unit were excellent. They made us feel that they had all the time in the world for us and, although we had to terminate because the chances of our baby surviving was zero, we never felt pitied, but extremely supported – a real skill.'*

Formerly pregnant woman

Screening programmes are different from normal clinical care because they offer tests to people who may not perceive themselves to have or be at risk of the condition in question. It is therefore important that everyone from policy-maker to practitioner appreciates that the individual patient, or client's needs are the central focus – they must be well informed and able to take decisions for themselves about whether or not to accept the offer of screening tests. This is reflected in the *National Service Framework for Children, Young People and Maternity Services* (DH, 2005):

*'Each pregnancy is different and each woman has different social, physical and emotional needs as well as specific clinical factors that may affect her pregnancy. Good maternity services place the mother and her baby at the centre of care, and plan and provide services to meet their needs.'*

This is the philosophy for the whole *Screening Choices: A resource for health professionals offering antenatal and newborn care* programme.

Even if you are confident that you understand the issues and information you need to discuss with parents in relation to antenatal and newborn screening, enabling them to make informed choices is not always straightforward. Individuals have different needs for information and support that can depend on their personality and capacity to understand often complex information, as well as a range of other factors. One of the standards in the *National Service Framework for Children, Young People and Maternity Services* (DH, 2005) requires all maternity services to ensure that: *'Women have access to supportive, high quality maternity services, designed around their individual needs and those of their babies.'*

Activity 8 will help you to reflect on how individual factors may affect different people's needs in relation to screening. The advanced activity asks you to analyse your skills in recognising and meeting these needs.

### Activity 8

## Meeting individual needs

#### Core

In your experience, what factors can affect the support and information parents need and their ability to make informed choices? Write down as many as you can think of.

#### Advanced

Reflect on three recent consultations about antenatal and/or newborn screening. How successfully were you able to tailor the consultations to meet the individual needs of the women and families concerned? Have there been occasions when you felt you did not meet individual needs as well as you should? Why do you think this was?



Use your workbook  
to do this activity

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## Feedback

### Core

Many factors can affect the choices parents make about antenatal and newborn screening including:

- Their religious/moral beliefs
- Their family origin or culture
- Their social/economic situation
- Their beliefs about disability.

Other factors affect their ability to make informed choices about screening, including:

- Their ability to understand/interpret the information they are given
- Problems such as dependence on drugs/alcohol
- Their ability to understand the information provided to them
- Whether they are in a position to act on informed choice (eg, if they are in prison, or others have significant influence over them).

These and other factors can influence their needs for information and support from you or others in your service. For example, they may need additional resources such as translation services or written information in another language or in Braille, extra help to understand information or additional information that is specific to their situation. Do you think your service is flexible enough to meet a wide range of individual needs?

### Advanced

In reflecting on the recent consultations you may have come across instances where you were particularly successful in meeting women's individual needs. If so, it is helpful to analyse why you were successful. Did you have access to a useful resource, or use a technique that was particularly effective? Could you use this reflection to help you in future consultations?

If you identified occasions when you were not as successful as you would wish, try to work out why. Can you learn anything from this to improve your practice in future consultations? Have you shared this with your colleagues? If not, you may want to raise it in a team meeting or during clinical supervision.

*Informed choice for everyone: valuing diversity* provides an opportunity to reflect on how your own assumptions, values, beliefs and attitudes may influence how you work with women and their families. It aims to help you to ensure they are all enabled to make informed choices about antenatal and newborn screening.

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Having studied this unit you should have developed an understanding of screening in general, and how it relates to antenatal and newborn screening. You should also appreciate the importance of understanding the screening programmes in which you are involved and being able to discuss these with pregnant women and new mothers. Activity 9 is another skills assessment that should help you to check whether you have met your learning outcomes and identify any areas where you may need to increase your skills or knowledge. Other units in this programme, or materials from the other antenatal and newborn screening programmes may help you with this.

## Activity 9



Use your workbook to do this activity

## Self-assessment

Look at the seven statements below and in your workbook rank how much you know about each, and how competent you feel about completing the task(s) involved in them. Use a scale of 1–5 (1 being NOT very competent and 5 being VERY competent). Compare your scores with the self-assessment you did before studying the unit.

1. I can discuss the purposes of screening programmes and how they can be realised.
2. I can explain what is meant by the term 'screening' and the concepts of screening and diagnostic tests.
3. I can define the following technical terms used in screening programmes easily and in a way that patients, clients and others can understand:
  - Sensitivity
  - Specificity
  - False positive
  - False negative
  - Detection rate.
4. I can find information on:
  - Current national programmes
  - Local programmes and arrangements
  - Specific screening programmes.
5. I understand the importance of quality assurance in screening programmes and the organisation involved in ensuring this.
6. I can describe a screening programme in the context of antenatal and newborn care.
7. I can explain my own role within an antenatal or newborn screening programme.

### What now?

If you feel you still need to improve your skills and knowledge you may want to look at the additional resources listed in the Toolbox. Alternatively, you may need to study materials provided by the screening programmes, or other units within this programme.

## Focus activity



Use your workbook  
to do this activity

## The case for screening

This focus activity is designed to help you apply what you have learned by working through the unit to situations you might encounter in clinical practice.

### Case example

Angela has two children aged 8 and 10. She is pregnant for the third time, and has been offered screening for sickle cell and thalassaemia and for Down's syndrome. This is the first time she has been offered screening for these conditions so she is anxious to know why.

How would you explain to Angela why she is being offered these tests?

### Think about:

- Developments in universal screening
- The potential benefits to her
- The limitations of screening
- The difference between screening tests and diagnostic tests.

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## Websites

- Cystic Fibrosis Trust: <http://www.cftrust.org.uk/>
- Department of Health Standards to Support Antenatal Screening for Infectious Diseases: <http://www.dh.gov.uk/assetRoot/04/06/61/91/04066191.pdf>
- National Society for Phenylketonuria: <http://www.nspku.org/>

## Sources of further information

- Abramsky, L., Chapple, J. (2003) *Prenatal Diagnosis: The human side* (2nd edn). Cheltenham: Nelson Thornes.
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- Wald, N., Leck, I. (2000) *Antenatal and Neonatal Screening (2nd edn)*. Oxford: Oxford University Press.

## The National Screening Programmes

- Cervical cancer: <http://www.cancerscreening.nhs.uk/cervical/>
- Down's syndrome: <http://www.nelh.nhs.uk/screening/dssp/home.htm>
- Fetal anomalies: <http://www.nelh.nhs.uk/screening/fasp/home.htm>
- Infectious diseases: <http://www.nelh.nhs.uk/screening/cpd/infectious.htm>
- National Screening Committee: <http://www.nsc.nhs.uk>
- National electronic Library for Health Screening Specialist Library: <http://rms.nelh.nhs.uk/screening>
- Newborn blood spot: <http://www.newbornscreening-bloodspot.org.uk>
- Newborn hearing: <http://www.nhsp.info/index.shtml>
- Sickle cell and thalassaemia: <http://www.kcl-phs.org.uk/haemscreening>

## Standards and Guidance

CDR Weekly (2003) Screening for infectious diseases in pregnancy: Standards to support the UK antenatal screening programme. *CDR Weekly*; **13**: 3.

National Institute for Clinical Excellence (2003) *Antenatal Care: Routine care for the healthy pregnant woman*. London: NICE.

Department of Health (2004) *Model of Best Practice for Down's Syndrome Screening Services*. London: DH.

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<http://www.nelh.nhs.uk/screening/dssp/standards.htm>

UK Newborn Screening Programme Centre (2005) *Newborn Blood Spot Screening in the UK*. <http://www.newbornscreening-bloodspot.org.uk>

Unit: Screening in antenatal and newborn care

# Certificate of completion

This certificate acknowledges that I have worked through the above unit from the Screening Choices programme at Core/Advanced\* level, and I have achieved its learning outcomes.

I am able to:

- Discuss the purposes of screening programmes and how they can be realised
- Explain what is meant by the term screening and the concepts of 'screening' and 'diagnostic' tests
- Define the following technical terms used in screening programmes: sensitivity, specificity, false positive, false negative, detection rate
- Find information on: the current national programmes; local programmes and arrangements; and specific screening programmes
- Understand the importance of quality assurance in screening programmes and the organisation involved in ensuring this
- Describe a screening programme in the context of antenatal and newborn care
- Explain my own role within an antenatal or newborn screening programme.

I attach evidence of my learning and confirm that this is a result of my own endeavours and fully acknowledges the work of others.

Signature

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Name

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Job title

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Date

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\* Delete as appropriate

# Factsheet:

## Screening in antenatal and newborn care

### Antenatal and newborn screening programmes

This Factsheet accompanies *Screening in antenatal and newborn care*, and contains a selection of reference information on antenatal and newborn screening. Further information on specific screening programmes is available from the training materials produced to accompany these programmes and from the programme websites listed in the Toolbox.

The National Institute for Clinical Excellence (NICE, 2003) has developed a clinical guideline on *Routine Care for the Healthy Pregnant Woman*, which sets out which antenatal tests women should be offered and at what stage of gestation.

#### Before 16 weeks

Women should be offered:

- Blood tests for
  - blood group, rhesus status and red cell antibodies (this is offered again at 28 weeks to women with no antibodies on initial testing)
  - haemoglobin disorders (sickle cell disorders and thalassaemia)
  - haemoglobin level (for anaemia)
  - hepatitis B virus
  - HIV
  - rubella susceptibility
  - syphilis serology.
- A urine test to screen for asymptomatic bacteriuria.
- An ultrasound scan to determine gestational age, identify the number of fetuses and confirm fetal viability.

All women should also be offered screening to detect risk of Down's syndrome. A number of tests can be used alone or in combination (see box on page 31), and the tests used dictate when they are performed. The following tests meet the NICE and UK National Screening Committee (NSC) standard of detecting at least 60% of affected pregnancies and having less than 5% false positive results (70% and 3% respectively by 2007):

- From **11 to 14 weeks**
  - nuchal translucency (NT)
  - the combined test (NT, hCG and PAPP-A)
- From **14 to 20 weeks**
  - the triple test (hCG, AFP and uE3)
  - the quadruple test (hCG, AFP, uE3 and inhibin A)
- From **11 to 14 and 14 to 20 weeks**
  - the integrated test (NT, PAPP-A + hCG, AFP, uE3, inhibin A)
  - the serum integrated test (PAPP-A + hCG, AFP, uE3, inhibin A).

Women whose initial screening indicates an increased risk of Down's syndrome should be offered a diagnostic test. Most services offer one of the following tests, which are invasive and carry a slightly increased risk of miscarriage:

- Amniocentesis
- Chorionic villus sampling
- Cordiocentesis.

#### Between 18 and 20 weeks

Women should be offered another ultrasound scan, to detect structural anomalies.

### **Down's syndrome screening tests**

Nuchal translucency (NT) uses ultrasound to measure the amount of fluid at the nape of the fetal neck. An increased amount of fluid may indicate that the fetus has Down's syndrome or another chromosomal, structural, or genetic anomaly.

Biochemical markers that indicate an increased risk of Down's syndrome can also be detected by screening tests:

- Human chorionic gonadotrophin (hCG)
- Pregnancy-associated plasma protein A (PAPP-A)
- Alpha-fetoprotein (AFP)
- Unconjugated oestriol uE3
- Dimeric inhibin A.

### **Diagnostic tests**

These are also used to detect sickle cell disorders, thalassaemia and a range of other conditions.

Amniocentesis is performed by inserting a fine needle through the abdominal wall into the uterus and withdrawing a small amount of fluid from the sac surrounding the fetus.

Chorionic villus sampling involves inserting a fine needle through the abdominal wall or a thin plastic tube through the vagina and cervix to take a tissue sample from the placenta.

Cordocentesis involves taking a blood sample from the umbilical cord. It is usually only carried out in emergency situations from 18 weeks gestation, when a quick answer is required.

## **Other tests**

### **Blood group**

*This is a diagnostic test*

Testing to identify blood group has been offered to pregnant women in the UK for about 30 years in case the woman needs a blood transfusion.

### **Rhesus status and red cell antibodies**

*This is a screening test*

This test has two functions – to determine maternal rhesus status and to identify the presence of, or assess the type and amount of red cell antibodies, which can cause rhesus incompatibility between mother and baby.

### **Sickle cell disorders and thalassaemia**

*This is a screening test*

Haemoglobin disorders such as sickle cell and thalassaemia are inherited, and prevalence varies among different population groups.

Sickle cell disorders are a range of genetic conditions that cause the bone marrow to produce red blood cells which become sickle-shaped when deoxygenated. These are broken down by the body very quickly, leading to anaemia. They can also block small blood vessels, causing severe pain and possibly organ damage, and individuals with these conditions are more prone to overwhelming bacterial infections.

Inheritance of sickle cell disorder depends on the sickle cell or haemoglobin variant carrier status of both parents. The baby will not inherit it unless both parents have the disorder, or are unaffected carriers, or one is a carrier and the other has another type of unusual red blood cells – in which case they are unaffected and may be unaware of their status, but can have affected children. If the mother is found to be a carrier of sickle cell,

the father can be screened to calculate the risk to the baby. *More information on inheritance is contained in the unit **Understanding genetics**, and in the training for the sickle cell and thalassaemia screening programme.*

It is estimated that 160 babies are born each year in England with a sickle cell disorder. Prevalence of sickle cell carrier rates is:

- Black African populations 10–28%
- Black Caribbean populations 4–11%
- Cypriot populations 0.75%
- Indian populations 1%
- Northern European populations 0.05% (Davies et al, 2000).

Beta thalassaemia major causes severe anaemia in infancy, which is usually fatal by 10 years if untreated. It is inherited in the same way as sickle cell disorders and tested for and diagnosed in the same way. On average, 17 babies are born each year with thalassaemia. Its prevalence of carriers among ethnic groups is:

- Bangladeshi populations 3%
- Black African and Caribbean populations 0.9%
- Chinese populations 3%
- Cypriot populations 16%
- Indian populations 3.5%
- Northern European populations 0.01%
- Pakistani populations 4.5% (Davies et al, 2000).

Antenatal screening for sickle cell disorders and thalassaemia aims to identify women with sickle cell or thalassaemia disorders so that they can receive appropriate care, as well as babies at risk in early pregnancy so that their parents can be offered genetic counselling. This can help them to decide whether or not to have amniocentesis or cvs, which are diagnostic tests. If their baby has a sickle cell disorder or thalassaemia, they can then decide whether to continue with their pregnancy.

*Specific training is available on screening for sickle cell disorders and thalassaemia (<http://www.kcl-phs.org.uk/haemscreening>).*

## **Anaemia**

*This is a diagnostic test*

In pregnancy, low haemoglobin (Hb) levels in the blood are the most common cause of iron deficiency. Pregnancy and the increase in overall circulating blood volume means mothers need more iron than usual. Anaemia can cause dizziness, fatigue and general ill-health, and in severe cases can lead to low birth-weight babies.

Haemoglobin levels vary through pregnancy, so it is recommended that they are checked at various stages. Up to 12 weeks the level should be at or above 11g/dl and at 28–30 weeks it should be at or above 10.5g/dl. Where there is a suspicion of anaemia, serum ferritin testing is recommended as it is more sensitive and specific in detecting iron stores. If necessary, the woman can be offered iron supplements, but should also be advised about eating iron-rich foods.

## **Infectious diseases**

Testing for infectious diseases enables women to receive treatment to reduce the risk to themselves and their babies:

- Asymptomatic bacteriuria (urinary tract infection) increases the risk of pyelonephritis (kidney infection) and preterm birth. It can be treated with antibiotics. *This is a diagnostic test.*
- Some people are asymptomatic carriers of hepatitis B virus, which can be transmitted from mother to child. If the mother is infected, the child can be vaccinated soon after birth to prevent long-term liver damage. *This is a diagnostic test.*
- HIV can also be transmitted from mother to child. Testing for HIV means the mother

can receive antiviral drug treatment to maintain her health and reduce the risk to her baby. The risks to the baby can also be reduced by a caesarean delivery and bottle-feeding rather than breast-feeding. *This is a diagnostic test.*

- Rubella (German measles) is a mild, often asymptomatic disease. However, in pregnancy increases the baby's risk of blindness or deafness, heart defects and learning disabilities. Screening identifies whether the woman has a current infection or low immunity to rubella. The purpose of screening the pregnant population is to identify women who lack rubella specific antibody and ensure that the offer of rubella vaccine is made in the postnatal period. This would give a protection for future pregnancies. The consequences of exposure of the fetus to primary rubella infection during pregnancy can be catastrophic, the risk being greatest in the first 16 weeks of pregnancy. An antibody level of less than 10iu/ml is considered to be 1gG negative. Results should be reported as Rubella 1gG 'detected' (immune) or 'not detected' (non-immune).

All 1gG negative women should be offered MMR vaccine in the immediate postnatal period and advised not to conceive within one month of MMR vaccination. *This is a diagnostic test.*

- Syphilis is a sexually transmitted disease, but can be passed from mother to child, when it can cause serious physical problems and learning disabilities and increases the risk of stillbirth. The disease can be treated with antibiotics with no risk to the baby. *This is a diagnostic test.*

### **Fetal anomalies**

The object of fetal anomaly ultrasound screening is to identify:

- Anomalies that are not compatible with life
- Anomalies associated with high morbidity and long-term disability
- Fetal conditions with the potential for intrauterine therapy
- Fetal conditions that will require postnatal investigation or treatment.

A number of minor anomalies are also picked up such as cleft lip and palette. Ultrasound has been found to detect 44.7% of all fetal anomalies at the routine 18–20-week scan, although detection rates vary with different anomalies. Where a routine scan suggests an anomaly depending on its nature a more detailed scan or diagnostic testing may be indicated for confirmation. *This is a screening test, although in cases where it identifies anomalies for which there are no further tests it is considered to be a diagnostic test.*

### **Down's syndrome**

Down's syndrome is a chromosomal disorder that causes learning disabilities, although it is also associated with other problems such as heart defects, leukaemia, epilepsy and Alzheimer's disease. The incidence of Down's syndrome in England and Wales was 6.2/10,000 live births in England and Wales in 1998.

Screening estimates the risk of a pregnancy being affected by Down's syndrome. Women whose pregnancies are found to be at a risk greater than one in 250 can be offered diagnostic testing (amniocentesis or chorionic villus sampling). This enables them to choose whether or not to continue with their pregnancy. Those with affected babies who do choose to continue can be given help and support to prepare for their baby's special needs.

*Specific training is available on screening for Down's syndrome (<http://www.nelh.nhs.uk/screening/dssp/home.htm>)*

*More information on Down's syndrome is available from the Down's Syndrome Association: <http://www.downs-syndrome.org.uk>*

## Newborn screening

A number of screening tests are offered within the first 10 days of the baby's life.

- A physical examination focuses on the baby's vision, hips and heart. Any problems detected may be correctable with surgery or other treatment, or the child may need to be referred to specialist services to help the child and family to deal with the challenges that any detected conditions may present.
- A hearing test is offered so that the child can be referred to specialist services if necessary.

A blood spot test is also offered to all newborn babies to screen for:

- Phenylketonuria (PKU), a genetic disorder which prevents the child processing phenylalanine, an amino acid found in protein food. This disorder can lead to brain damage unless the child is put on a strict, usually, lifelong diet that is low in phenylalanine. This requires close supervision by a dietitian or appropriately qualified doctor, and the cooperation of the child and family.
- Congenital hypothyroidism (CHT), a condition in which the thyroid gland does not exist or produces too little thyroid hormone (thyroxine). It may lead to abnormal growth or development and slower mental function. It can be treated with oral thyroxine, which must be taken daily for life.
- Cystic fibrosis (CF), a genetic disease that affects a number of organs (especially the lungs and pancreas) by clogging them with mucus. It is most common in white European people, affecting 1:2,500. About 2–5% of white people carry the CF gene. Until the 1950s only half of those born with CF lived beyond two-and-a-half years of age (Farrell and Mischler, 1992). Improved treatment has extended their life expectancy to an average of 31 years and improved their quality of life. Treatment includes antibiotics for respiratory infections, vitamin supplements, inhaled bronchodilator drugs and physiotherapy to keep the lungs clear of mucus.

*More information on cystic fibrosis is available from the blood spot screening programme (<http://www.newbornscreening-bloodspot.org.uk>) and from the Cystic Fibrosis Trust: <http://www.cftrust.org.uk>*

- Medium chain acyl-CoA dehydrogenase deficiency (MCADD), an autosomal recessive genetic metabolic condition that makes it difficult for the body to break down fatty acids to produce energy. It can cause hypoglycaemia, fatigue, lethargy, vomiting, seizures, coma and even sudden death. Treatment involves preventing metabolic crisis by avoiding fasting and close monitoring to determine safe periods between meals. A high carbohydrate emergency regimen must be initiated if the child is unwell. A pilot project to evaluate the benefit of screening for MCADD is being undertaken in some areas of England.
- Sickle cell disorders, a group of inherited conditions which may result in complications such as overwhelming bacterial infections, acute and chronic anaemia, severe painful episodes called 'crises', and death. Approximately 1:2500 births are affected by sickle cell disorders. Screening for these conditions is currently being incorporated into the blood spot screening programme.

## Factors affecting NSC selection of screening programmes

The UK National Screening Committee uses a range of criteria to decide which conditions should be the subject of screening programmes (<http://www.nsc.nhs.uk>). These are paraphrased and explained below.

### The condition

- The condition should be acknowledged as an important health problem, in terms of its impact on both the people affected (and/or their families) and on society more widely. *A test that detects a condition that is serious but only affects one baby in 500,000 would not be offered to all pregnant women as it would not be an effective use of resources.*

*Equally, screening would not be offered to detect a common condition that does not cause serious problems for those affected.*

- The condition should be well understood and either the condition or a risk factor should be detectable.  
*It is of little benefit to detect conditions that are poorly understood or to try to detect conditions for which there is no reliable test.*
- If the condition can be prevented or the risk reduced, all the practical steps to do this should have been taken.  
*It is far better to prevent a condition if this is possible, than to let it develop then treat it.*
- It is also important to understand the physical and psychological implications for affected people and their families of the condition screened for.  
*Women, partners and families need to be given access to the appropriate information, treatment and support, so professionals must understand their likely needs when they receive their results.*

### **The test**

- There should be a simple, safe, precise and validated screening tool, the normal range of results should be known and a cut-off level for increased risk (or diagnosis of the condition) defined and agreed.  
*If a test could give results of 1–10 and the normal range is defined as 1–6, people with a score of 7 or more will be considered at increased risk or to have the condition.*
- The test should be acceptable to those who undergo it, although this does not necessarily mean it must be simple and painless.  
*A test may cause pain or discomfort, but if a woman accepting it for herself or her baby believes this is worth enduring in order to gain more information, she will still accept the test.*
- There should be an agreed policy on what to do in situations where the test is positive, and on the choices available to individuals.  
*This may involve, for example, referral to specialist services or for further tests.*
- If the test is to detect alterations (such as fetal anomalies), there should be clear criteria on which are covered by screening if it does not cover all possible alterations.  
*A huge number of alterations may be detectable by screening. However, screening resources are limited so services need to agree which tests are to be offered to all women or babies. The criteria may differ between services; for example in areas of high prevalence all pregnant women are offered screening for sickle cell disorders and thalassaemia, while in areas of low prevalence this may not be the case.*

### **The treatment**

- There should be an effective treatment or intervention for patients identified through early detection, with evidence that this leads to better outcomes than late treatment.  
*There is little point in screening for a condition if nothing can be done once the results are received.*
- There should also be agreed, evidence-based policies on who should be offered treatment or interventions, and what they should be offered.  
*Some screening may detect conditions that affect some people more severely than others, or a number of treatments and interventions may be available. Professionals need clear guidelines on who to treat and how to treat them.*
- Finally, before healthcare providers participate in screening programmes, the clinical care they offer to people requiring it should be of an agreed standard.  
*There is little point in running a screening service if people cannot be offered high*

quality services when they receive their results.

### **The screening programme**

- There should be high-quality research evidence that the programme reduces morbidity or mortality.  
*For example, although research evidence on the benefits of newborn screening for CF is inconclusive, a number of case studies suggest that children who are diagnosed after newborn screening might be healthier than those diagnosed later.*
- If the test is aimed solely at giving people information to allow them to make an informed choice (such as whether to continue with a pregnancy), there should be evidence that the test accurately measures risk. It is also important that the person is given good information in a form that they can understand.  
*People should not be asked to make decisions about termination based on unreliable tests, or if they do not understand the issues.*
- There should be evidence that the whole programme, from testing and diagnostic procedures to treatment or intervention, is generally acceptable to health professionals and the public.  
*For example, although some people disagree with all abortions, there is a general acceptance of terminations where babies have serious conditions that cannot be treated.*
- Where screening is for a genetic alteration, the programme should also be acceptable to people identified as carriers and to other family members.  
*For example, screening for sickle cell disorders is generally accepted as a good thing by people who have the condition. However, people with other conditions, such as cleft lip, may find it unacceptable to screen specifically for the condition antenatally because it suggests parents may want to consider terminating an affected fetus.*
- The benefit from screening should outweigh the harm caused by the test, diagnostic procedure and intervention or treatment.  
*For example, if a test detected a serious condition but resulted in a 50% miscarriage rate, this may be considered unacceptable, as might a treatment which controlled a condition but had serious side-effects.*
- Screening must provide value for money.  
*This means the cost of the programme must be balanced in relation to all expenditure on the condition if it is not detected.*
- There should be an agreed plan for managing and monitoring screening programmes, and an agreed set of quality assurance standards against which they can be assessed. Adequate staffing and resources must also be in place.  
*Even with careful planning, there is no guarantee that screening programmes will be operated effectively. They should, therefore, be managed and monitored, and assessed against an agreed set of standards. There should also be enough staff and resources to operate the programme.*
- Before a screening programme is set up, all other options for managing the condition should have been considered, such as improved treatment or providing other services.  
*Screening must be the best way of dealing with the condition.*
- Professionals must have access to evidence-based information that explains the consequences of testing, investigation and treatment.  
*It is not enough to simply expect professionals involved in screening to ensure people can make informed choices – they need training and information resources to help them in this.*
- Finally, public pressure to widen the availability of screening, reduce the screening interval or increase the sensitivity of testing should be anticipated. The decisions made about these should be scientifically justifiable to the public.  
*People may argue that other conditions should be screened for, that screening should be done more regularly, or that a more sensitive test should be used. There should be clear scientific arguments to support the current situation.*

## References

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