Part One
What is Screening?

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 from about 80–90% to 6–8% the chance of neurological impairment in babies born with the condition of cases (Health Technology Assessment, 1997)

- Incidence of cervical cancer in Scotland fell by 48.8% between the baseline year 1986 and 2007. This fall is directly related to the cervical cancer screening programme (Scottish Cervical Screening Programme ISD 2010). Cervical screening is thought to prevent around 1,300 deaths and up to 3,900 cases of cervical cancer per year in the UK

When people decide whether to accept or decline screening 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. To facilitate this, you need to be able to offer them the information, advice and support they need in order to enable women, their partners and families to make informed choices about offers of pregnancy and newborn screening. You also need to understand the concept of screening in general. This is quite complex – here is 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
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General Principles of a Screening Programme

a) **The conditions screened for should pose an important health problem.**
There would be no support for setting up a screening programme for anything that was not deemed to be an important health problem.

b) **The natural history of the condition should be well understood.**


c) **There should be a recognisable latent or early stage.**
Early detection offers more options for treatment and preventative health care for those affected.

d) **Treatment of the problem at an early stage should be of more benefit than treatment started at a later stage.**


e) **There should be a suitable test or examination.**
The decision about which screening method to employ is determined by its ability to detect a high proportion of cases at a stage where prognosis can be improved by early treatment. The criteria for assessing screening methods are that they should be:
- Simple
- Easy to apply
- Have a high sensitivity and specificity. Sensitivity is the ability of a test to detect a health problem in all individuals in whom it is present. Specificity is the ability to exclude those who don’t have the disease.
- Reproducible
- Cost effective
- Have a low risk to benefit ratio

f) **The tests or examinations should be acceptable to the population.**
Acceptability or otherwise of a screening programme is very much a personal decision. Education about the screening programme is therefore an essential factor in encouraging women/parents to engage in the programme.

g) **There should be adequate facilities available for the diagnosis and treatment of any abnormalities detected.**
It is extremely important when organising a screening programme that adequate facilities for diagnosis and follow-up are readily available.

h) **The chance of physical or psychological harm should be less than the chance of benefit.**
It is important that health professionals are aware of the advantages and disadvantages of screening. Screening has important ethical differences from clinical practice as NHS Scotland is targeting apparently healthy babies/women and offering to help make more informed choices about their health. There are uncertainties involved and it is important that women/parents have realistic expectations of what a screening programme can deliver. In any screening programme there is an irreducible minimum of false positive results (unaffected women/babies reported as being at increased risk of the condition) and false negative results (affected women/babies reported as not having the condition).

i) **The cost of case finding including diagnosis and subsequent treatment should be economically balanced against the benefits it provides.**
Recent studies seem to suggest that screening does not incur greater costs than standard diagnostic methods. It is essential that screening must be incorporated in to good follow-up and treatment programmes to achieve the potential benefits of early diagnosis.

Some of the conditions for which 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.

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

**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 then combined with a nuchal translucency (NT) measurement by ultrasound in order to generate a ‘combined’ risk assessment. Women in higher-risk categories are then offered a diagnostic test that
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involves examination of the baby’s chromosomes (karyotype) to look for the extra chromosome 21 that is present in Down’s syndrome.

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. Here is 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 chance 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 pregnancy care are associated with a small chance 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 chance 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.
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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. 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 chance; 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: pregnancy screening is offered to pregnant women when they attend an pregnancy 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.)
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• Ensure fail-safe follow-up of positive tests including arrangements for subsequent diagnostic testing.
  (Example: women with a higher chance of having a baby with Down’s syndrome, sickle cell disorder or thalassaemia can be offered amniocentesis or CVS.)

• 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 chance of mother-to-baby transmission.)

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.sct.screening.nhs.uk

Quality Assurance

Quality assurance and monitoring are essential components of any screening programme. Quality assurance can be defined by describing two of its main objectives:

• Guaranteeing minimum standards – developed and evaluated through NHS Quality Improvements Scotland (NHS QIS);
• The continual pursuit of excellence to reach more challenging but achievable standards.

A quality assurance programme covers several areas including:

• uptake
• consumer satisfaction – minimising anxiety, maximising acceptability;
• maximising the detection rate (sensitivity);
• minimising the false positive/false negative rate (specificity);
• the quality of equipment used to ensure a reliable diagnosis;
• the reliability and effectiveness of information systems;
• the quality and effectiveness of the management of the service;
• making information available in a format that takes account of the physical, cultural and mental health needs of the individual;
• the quality and effectiveness of local services including audiology, education, social work and speech and language therapy

To achieve this all personnel involved in pregnancy and newborn screening programmes must understand the concepts, goals and their own specific role in reaching these goals.

Setting Quality Standards

• Quality standards must define which factors to monitor, in order to develop and improve the service.
• Quality standards must be achievable and monitored carefully for them to have any value in the programme.
• The need for developments and improvements identified by an evaluation and monitoring procedure must be addressed.
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 chance 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 pregnancy 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 chance of a serious and untreatable condition. However, while newborn screening may not seem to pose the same difficulties as pregnancy 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.

Individual needs

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. Even if you are confident that you understand the issues and information you need to discuss with parents in relation to

Pregnancy and newborn screening

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

Pregnancy screening

Pregnancy 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 chance of, conditions such as neural tube defects, sickle cell disorders and thalassaemia.

In some cases, finding out that a fetus has a particular condition allows therapy to be offered during pregnancy 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 pregnancy screening identifies an increased chance 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.

Women are offered one or more screening tests which are used to give an estimate of their baby’s chance 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 these tests are invasive and carry a small chance of miscarriage.
pregnancy 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.

Many factors can affect the choices parents make about pregnancy 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
- Whether they are in a position to act on informed choice (e.g., 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, such as easy read versions, that is specific to their situation.

**Conclusion**

You should now be able to:

1. Discuss the purposes of screening programmes and how they can be realised.
2. Explain what is meant by the term ‘screening’ and the concepts of screening and diagnostic tests.
3. 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. Describe a screening programme in the context of pregnancy and newborn care.
5. Explain your own role within a pregnancy or newborn screening programme.
Perceptions of risk/chance

Pregnancy and newborn screening programmes aim to benefit women and their families by giving them information about their own health, or that of their fetus or baby that can enable them to make informed choices about treatment or other options. However, a range of risk is associated with screening and/or diagnostic tests and the implications of their results.

All the women, partners and families you work with will react differently to these risks depending on their perception of them. This will affect the way they try to come to decisions and what those decisions are. People’s perceptions can be influenced by their capacity to understand the information, and other factors that are individual to them and their own circumstances.

Two women with exactly the same results, explained in exactly the same way, will not necessarily perceive their risk in the same way (Marteau, 1999). For example, a woman with a 25% chance of having a baby with a sickle cell disorder may not see this in the same way if a member of her family is mildly affected by the condition in question as another woman who has a severely affected relative.

Equally, a woman who has decided that she would prefer to end a pregnancy if her baby was found to have Down’s syndrome may see a one in 35 chance very differently to one who has decided to continue with her pregnancy even if it is affected. The way people perceive risks and make decisions is also affected by the fact that the demands of daily life mean they often have limited time to make decisions of all kinds.

In order to cope with this, they use a number of ‘decision rules’, known as heuristics, which help to
provide quick answers. Three common heuristics are:

- **Availability** – people judge things to be more likely to happen if they find them easy to imagine or think of an example from their own experience. For example, someone who works with children with a particular condition may perceive their own baby’s chance to be high, regardless of information such as numbers and percentages they may be given.

- **Representativeness** – people judge the probability of an event by finding a ‘comparable known’ event and assuming the probabilities will be similar. For example, a woman who has had a pregnancy in which a serious fetal anomaly was detected may assume her next will have the same problem, regardless of whether or not her pregnancy is considered to be at higher risk.

- **Anchoring and adjustment** – people use an initial piece of information as an ‘anchor’ and use other information to adjust this. For example, a woman who is told that she has a one in 35 chance of giving birth to a baby with Down’s syndrome may see this as an acceptable risk until she is told that this means she is classified as being in the higher risk category.

These individual variations in the way women and partners perceive risks related to pregnancy and newborn screening highlight the importance of ensuring discussions about risk are a two-way exchange of information and ideas. Simply providing information on risk without discussing their specific circumstances and perceptions will not help women and their partners to make informed choices.

A wide range of factors may influence the way women and their partners perceive risk:

- Previous personal experience
- Their own physical and/or mental health
- Experience of illness among family members
- Obstetric history
- Cultural or religious values
- Personal attitudes to and understanding of chance, fate and inheritance
- Mood
- Age
- Controllability of chance and coping with uncertainty
- Previous health service experience.

**We would always recommend using the word ‘chance’ when discussing risk with women and their families**

**Presentation of risks/chances**

While factors related to their own experiences and circumstances can influence the way women and their partners perceive risk related to pregnancy and newborn screening and the decisions they make as a result, another potentially major influence is the way health professionals’ present information about chance (Marteau, 1999). This is known as ‘framing risk’.

For example, being told ‘You have a one in 20 chance of having an affected baby may suggest a greater risk than ‘You have a 19 in 20 chance of having an unaffected baby’.

It is important to understand that expressions of risk often carry a positive or negative message, and the potential for this to influence decisions about screening.

You can also minimise the possibility that your framing of risk affects women and their partners’ decision-making by presenting information in a number of ways. So, rather than simply saying ‘You have a one in four chance of …’, you could continue ‘or put another way, a 25% chance’ you can also add ‘but that also means you have a three in four or 75% chance of …’.

Selective information-giving can also affect women and their partners’ ability to make informed choices about screening. You should therefore ensure you do not leave out information that may be important in their decision-making, such as details about conditions for which screening is being offered, or risks associated with diagnostic tests.

In order to avoid framing risks in a way that may influence women and their partners’ decisions, it is important to be aware of your own views on the issues you are discussing.
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It can be difficult to avoid showing your own views – particularly in your non-verbal communication.

For example:
• Do you think women who are carrying babies affected by Down's syndrome or sickle cell disorders should continue with their pregnancy?
• Do you feel women should only have diagnostic tests if they intend to terminate their pregnancy if it is affected?
• Do you feel that testing pregnant women for HIV is unnecessary if they are not in a higher-risk group?
• Do you feel that parents who decline newborn screening tests are irresponsible?

You may have strong beliefs about these and other issues related to pregnancy and newborn screening. However, your personal values and beliefs should not influence your professional practice when presenting information on screening options or results, or what information you choose to give.

It can be difficult to avoid showing your own views – particularly in your non-verbal communication. You may also subconsciously give clues about your own attitudes and feelings through the language you choose.

Here are some examples of statements that women may say to you and ways that could help them to make an informed choice (Adapted from UK NSC Antenatal Screening Choices http://cpd.screening.nhs.uk/screeningchoices)

1. ‘I don’t want any screening while I’m pregnant. My sister and friend had everything they were offered, and it just made them worry. Their babies were fine but the tests spoiled their pregnancies.’

You would need to ensure this woman has all the information she needs about the purposes of screening programmes, to ensure she understands the possible implications if her baby is affected by any of the conditions tested for in the pregnancy period. This could include information on incidence of particular conditions screened for, risk factors for them, what the tests involve and the effects of the conditions. You may provide written information for her to take away so that she can consider her decision further if she wishes.

2. ‘They only offer CVS when there’s a one in 250 or less chance of the baby having Down’s syndrome. They told me there’s a one in 850 chance of my baby having being affected – that’s still a chance, and I want to have a CVS so I know for sure.’

This woman may not understand the risk of miscarriage associated with CVS, and how this compares in numerical terms with the chance of her baby being affected with Down’s syndrome. However, she may feel so strongly that she could not cope with a baby with Down’s syndrome that she prefers to take the chance of having a CVS to give her a definitive answer. Your role would be to find the reasoning behind her statement. You would also have to ensure she understands how the risks of CVS and her baby being affected with Down’s syndrome compare, and that she has a clear understanding of Down’s syndrome. This will ensure her choice is fully informed. She may also find it helpful to contact an organisation like Pregnancy Results and Choices (http://www.arcuk.org), the Down’s syndrome Association (http://www.downd syndrome.org.uk) or Down’s syndrome Scotland: http://www.dsscotland.org.uk/

3. ‘I’ve cut down on my smoking, but I’m not giving up completely. I don’t see how five or six a day can do any harm.’

This woman may not fully understand the risks to the fetus of smoking during pregnancy, and the possible long-term health effects to her child when it is born. However, she may understand these risks and have tried but failed to stop smoking, or believe the potential effects on the fetus caused by the stress of not smoking would be as great. You would need to explore her understanding of the effects of smoking, and offer smoking cessation literature or sources of help such as the NHS Health Scotland ‘can stop smoking’ web site or local smoking cessation clinics. Involving her partner or other family members in helping her to stop smoking or further cut down her cigarette consumption may also be helpful, but ultimately it is her decision whether or not she stops smoking.
4. ‘If he has cystic fibrosis we’d rather not know until he gets ill. We’d only worry about it all the time.’

The benefits of screening for cystic fibrosis (CF) are less clear-cut than other conditions in the newborn blood spot screening programme – particularly PKU and congenital hypothyroidism (CHT), for which early treatment is important to prevent damage to babies’ development. However, your role would be to explain the potential benefits of early treatment and management of the condition, so that she can balance these against the possible anxiety that knowing her baby had CF would cause. You would also need to explore her understanding of CF – for example whether there was a family history of the condition (if this has not already been established during pregnancy care), or whether she has known someone affected by it, and whether she knows about treatment and management techniques.

5. ‘My mother-in-law said I shouldn’t have a diagnostic test for Down’s syndrome. She said we should see our baby as a gift from God and we shouldn’t do something that carries a chance of miscarriage.’

In some families, other members as well as the prospective parents are closely involved in decisions about issues such as pregnancy screening and diagnostic tests. You would need to explore whether this is acceptable to the woman and her partner, or whether they feel under pressure to make a choice that is not right for them. However, even if this is the case, they may feel the implications of disregarding the views of a senior family member are too great. There may also be religious reasons for them declining the test – knowledge of the teachings of religions prevalent in your local area may help you to know if this is the case.

If she is closely involved in decisions relating to the pregnancy, it may be helpful to involve the woman’s mother-in-law and ensure she fully understands the issues related to screening for Down’s syndrome.

6. ‘I don’t want them poking and prodding my baby. We just want to get her home – I’m her mother and I know she’s fine.’

Your role with these parents would be to ensure they understand all the conditions tested for in newborns, and the benefits of early identification. You should also ensure they understand what the tests involve, and the implications for their baby if conditions go undetected. If they still decline the tests it may be helpful to give them literature to take away, and to ensure that they know they can still have their baby tested if they change their minds in the future.

7. ‘My friend told me there are lots of conditions they can test for while you’re pregnant, but they only offer a few. I think that’s terrible – if a test is available, everyone should have the chance to have it.’

This woman may not understand the implications for the health service – and for women and families – of screening all pregnant women for all conditions for which tests are available. You would need to explain why screening programmes are set up for particular conditions. You would also need to explore why she has these concerns and whether they relate to any particular conditions. You may be able to reduce her anxiety, for example if she is worried about a genetic condition for which she has no family history.

**Risk/Chance in pregnancy and newborn screening**

The word ‘Risk’ and ‘chance’ are used in many different contexts and, as a professional, it is important for you to understand these differences and use the terms precisely. When risk or ‘chance’ comes into your consultations you must also realise that the women and families you work with will have their own understanding of the words and will use this and many other factors in their decision-making.

People use the word risk or chance in different ways. They talk about risks in everyday life: crossing the road, travelling in an aircraft, being in town late at night, or smoking. They may also discuss the risks of buying and selling shares on the stock market. In these examples, they may be referring to the likelihood that something harmful will happen, or to their perception of the potential seriousness of that harm (Marteau, 1999). You probably use these concepts of risk in your consultations with patients. For example,
Epidemiology

Information on the risks and benefits of screening is derived from epidemiological studies. For example, we know from national data that the incidence of PKU in babies born in the UK is 11 in every 100,000 (Smith et al, 1991). There are around 600,000 babies born in the UK each year, so this means on average 66 are born with PKU. Since we don’t know who these 66 will be, for any particular baby we can only say that the risk is about one in 10,000 or about 0.01% in the general population.

Researchers over the years have tried to make epidemiological information more useful by asking the question: What makes people more or less likely than average to get the disease in question? This can serve two main functions:

- From a research point of view epidemiology helps them to understand the causes of disease and how they could be prevented. A classic example of how epidemiology increases understanding of the cause of
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disease is that of smoking. In the 1950s Sir Richard Doll first reported that people with lung cancer were much more likely to smoke than people without the disease. Smoking was identified as a risk factor for lung cancer. It was then possible to look at harmful constituents in the smoke (‘carcinogens’) as potential causes of lung cancer. It also gives the impetus for measures to be put in place to reduce smoking in the population such as a ban on tobacco advertising or the introduction of smoking cessation services.

- In a practical way epidemiology helps services to target particular groups or individuals for prevention measures or treatment. Sickle cell disorders are more common in certain ethnic groups, so while all pregnant women are offered a screening test in high-prevalence areas in England, in areas of low prevalence screening, such as Scotland, screening involves finding out whether the woman is from one of the higher-risk groups. Being from a particular ethnic group is a risk factor for sickle cell disorder.

It is important to know about major risk factors related to the conditions screened for in pregnancy and newborn care (where they are known), and to be able to explain these to the women and families you work. You may also find that you are asked about the influence of particular risk factors. While you may not know these, you should know where you can find more information, or to whom you can refer them for more expert help. You should also understand how some risk factors may have a higher incidence in particular groups in society, and ensure you have the resources and support necessary to explain risk factors to different groups, such as those whose first language is not English.

### Absolute and relative risk

Epidemiologists talk about absolute risk and relative risk. These terms are useful in different situations, but if used inappropriately could cause misunderstanding and lead health professionals and service users to make ill-informed decisions. Absolute risk is the chance of developing a disease or condition over a particular time period. It answers the question:

What is my percentage risk of getting the disease?

The critical factor for interpreting absolute risk is the time dimension. A risk value is only meaningful if we know the time period over which it applies. For example, you might read in a newspaper that women who are 60 years old have a 2% chance of dying from heart disease.

Unless a time period is given this could mean anything from 2% of women die of heart disease as soon as they reach the age of 60 to 2% of women who reach the age of 60 will die of heart disease at some time from 60 onwards. To be meaningful, the statement would need to say that women who are 60 years old have a 2% chance of dying of heart disease within X years.

Relative risk is the incidence of a disease in an ‘at risk’ group divided by the incidence of the disease in those not ‘at risk’. (At-risk groups are those who possess risk factors associated with particular diseases, such as smoking, drug-taking, older age, particular ethnic group etc.) Relative risk answers the question How many times more likely are those ‘at risk’ to get the disease than those not ‘at risk’? It is useful when considering possible causes of the disease but is not usually useful in the context of discussions about the advantages and disadvantages of screening.

Be careful. It is easy to misunderstand different risks/chances, or to use them in a misleading way.

1. Around 250 babies of the 600,000 babies born in the UK this year will have sickle cell disorders. - This refers to the absolute risk of sickle cell disorder for all babies born in the UK this year.
2. Pregnant women aged 40 have a 15 times greater chance of having a baby with Down’s syndrome than those aged 20 (National Down’s syndrome Screening Programme, 2004) - This refers to a relative risk. The chance of having a pregnancy affected by Down’s syndrome is 1:1,500 in women aged 20, and 1:100 in women aged 40.
3. Children whose parents both carry the gene for cystic fibrosis are twice as likely to be unaffected carriers than they are to be affected with the condition (UKNSPC, 2005) - This refers to a relative risk – children born to parents who both carry the gene for CF have double the chance of being unaffected carriers as they do of having the condition.
4. Women with HIV infection have a 25–45% chance of passing the virus to their children during pregnancy, birth or when breastfeeding if they do not receive preventive treatment (Avert, 2005) - This refers to an absolute risk of vertical transmission of HIV from mother to child if mother and baby do not receive prophylactic treatment and the mother breastfeeds.

Part of your professional expertise is in weighing up whether a woman has risk factors for particular problems and this may come into some of your discussions in her pregnancy care and care of her newborn baby. You will, for example, certainly want to find out whether she, her partner or other close family members smoke, and give advice to stop smoking in order to protect their baby’s health. You might also enquire whether they have other risk factors such as drug-taking or a history of depression so that you can make sure that appropriate support is available. You will also be able to understand how you can discuss that chance in qualitative (risk factors) and quantitative epidemiological (incidence of disease, absolute risk, relative risk) terms.

Sometimes, although we can identify risk factors, we cannot use these to target our interventions. For example, although we know that some higher-risk groups such as intravenous drug users are at increased risk of HIV, we cannot just target these groups.

This is because cases in other groups may be missed or all those in higher-risk groups may not be identified, while the consequences of a missed diagnosis can be extremely severe for both mother and baby. Thus, for HIV a whole-population approach is recommended, in which all pregnant women are offered HIV screening, and based
on available evidence, midwives are urged to recommend HIV screening in each pregnancy.

### Risk/chance as probability

We have seen that professionals engage in ordinary discussions about risk or chance with their patients (the risk of miscarriage after amniocentesis or CVS), and also that ‘risks’ come into epidemiological terms with precise definitions – the absolute or relative risk of a particular disease, or condition. However, there is a third way in which we use ‘risk’, which is simply as another word for chance, probability or likelihood. Used in this way it is much the same as you might describe your chances of winning the lottery. The word ‘risk’ is also used in this way in epidemiology. But, in dealing with women and families, it is not always as simple this. Most people can probably remember standing in a line of 10 and wondering if they will be picked for the sports team (although in this case being picked is unlikely to be random). However, they may have more difficulty in visualising bigger numbers such as ‘one in 100’ or ‘one in 1,000’ or they may simply have difficulty in understanding what ‘one in 10’ means. So we need other ways of explaining it to them. The next section aims help you to communicate with women and families about risk or chance.

### Terminology and communication methods

When discussing pregnancy and newborn screening, you probably use a mixture of methods to help women and families understand their situation and make informed choices. These can include words, numbers, percentages, risk groupings and visual representations. Since different people understand and respond to different methods, Calman and Royston (1997) suggest that using a combination of several will improve understanding.

### Words related to risk

As already discussed, because of the way they use the word ‘risk’, most people will perceive the word in a negative way. This can affect the way they interpret what you have told them. Other words, such as ‘chance’, ‘possibility’, ‘probability’, ‘likelihood’, uncertainty and ‘danger’ may lead to different understandings – and hence feelings – about the same situation.

We would recommend always using the word ‘chance’ when discussing risk with women and their families.

When trying to communicate a degree of chance, it is likely that you use numbers, or percentages:

- ‘There is a one in five chance’
- ‘There is a 20% chance’

Some people may find it easier to understand one of these than the others, and it is good practice to use more than one and to find out if those you are working with have a preference. You should however, be consistent in your use of terminology. For example, you should not say ‘You have a one in 240 chance of having a baby with Down’s syndrome and a 1% chance of miscarriage if you have a diagnostic test.’

### Visual representations

Some people find it easier to understand concepts of risk or chance if visual aids are used to reinforce explanations. For example a 1% chance could be shown using 100 marbles in a bowl, with 99 of one colour and one of another, or by 99 circles of the same size and colour on a piece of paper and one triangle of a different colour. Other visual methods might include, pie charts, bar graphs, line graphs, tables or family trees.

It is important to choose the visual method that best fits the information you are trying to convey, and to ensure that visual representations give information in a neutral manner. For example, avoid using smiling and sad faces, or black and white icons in favour of icons of different shapes or of colours without ethnic associations.

### Risk groupings

The results of screening tests usually give a risk or chance of a condition rather than a diagnosis. They are often put into risk groupings, such as higher or lower risk or chance. These classifications can vary widely from one condition to another. For example, if a test for Down’s
syndrome shows a chance of one in 250 or less, it is judged to be in the higher-risk group. A woman’s increased chance of breast cancer due to family history however, is classified as low if it is around one in 10 because this is approximately the chance for the general population; breast cancer is a much more common condition than Down’s syndrome. However, these different classifications of similar sized risks can cause confusion and you should be careful when using these words.

As we have already discussed, different people find different communication methods helpful in understanding concepts such as risk or chance, so you may need to use more than one method when discussing issues of risk or chance associated with pregnancy and newborn screening. In the list of risks or chances and communication methods, the following matches would work:

- If parents’ carrier status for CF is unknown the most useful way to explain the risk is to use the overall prevalence in the UK of one in 2,500 live births. To make such a large number meaningful a community scale would probably be the most effective – for example you might put the risk in the context of the population of a local village with a population of around 7,500, saying that on average three would be likely to be affected.
- The age-related risk of Down’s syndrome can be expressed as a ratio or a percentage, and differs greatly between women aged 20 and those aged 45 or over (∗)
- Ideally, when cases of neural tube defects are reported, information will include whether or not the mother took folic acid (FA) before and/or during pregnancy. However, this information may not be available in all cases – either because the woman is unsure or the information is not available. A pie chart can illustrate the proportion of each possible group, showing how many of the women definitely took folic acid:
- Prevalence rates of haemoglobin disorders vary between different ethnic groups. However, as the numbers of mixed-race relationships increase, these prevalence rates are becoming less useful and meaningful. Also, if parents’ carrier status is known, the risk or chance of a pregnancy or newborn baby being affected can be calculated much more precisely than simply by describing prevalence in a particular group. Since the conditions are autosomal recessive, inheritance depends on the carrier status of both parents, so there are a number of possible permutations. Family trees can help to explain the risk of a pregnancy or baby being affected, unaffected or an unaffected carrier depending on the parents’ status.

## Conclusion

In summary, it is important to appreciate that communicating about chance is a two-way process (Edwards et al, 2002). It does not simply involve you giving information to women and their partners, but should be an exchange of information. This will help you to give information in the most meaningful way to ensure that they understand it.

You should now be able to:

1. Acknowledge individual variation in perception of risk/chance in both professionals and service users, give examples and understand the underlying factors.
2. Discuss the concept of presentation of risk/chance and the ways in which framing can influence women and their partners'/families' understanding.
3. Understand how epidemiology contributes to the understanding of chance in health and screening.
4. Explain absolute and relative risk and their relevance to the pregnancy and newborn screening programmes.
5. Discuss different uses of the term ‘risk’ eg, chance, probability, uncertainty.
6. Understand the different approaches to communicating the same risk/chance
7. Know the importance of using the word chance rather than risk when communicating with parents.
Part One
Understanding Genetics

Genetics is the study of how characteristics are passed on from one generation to the next (inherited) and how variation (differences between individuals) occurs.

Understanding of genetics has increased rapidly in recent years, particularly since the publication of the human genome – the ‘Book of Life’, which maps all the genes in the human body. The structure and function of genes provide a basis for the way in which the human body and all other organisms work and for the differences between them, including their experience of health and disease.

We have known for most of the last century about genetic diseases and the fact that some of these can be passed from one generation to the next – the hereditary disorders – but it is only recently with new technologies that we have been able to understand in more detail what alterations in basic genetic structure account for these conditions and to develop tests for them.

The UK National Screening Programme includes some genetic conditions both at the pregnancy stage (e.g. Down’s syndrome and sickle cell disorders) and for the newborn baby (e.g. phenylketonuria). This chapter will give you a basic understanding of genetics to help you explain these tests to patients, answer some of their questions and appreciate some of the difficulties they may encounter.

Scottish National screening programmes for genetic conditions

Pregnancy screening programmes
• Down’s syndrome
• Haemoglobin disorders - sickle cell disorders, thalassaemia disorders

Newborn screening programmes
• Congenital hip dislocation (also known as developmental dysplasia)
• Congenital hypothyroidism (CHT): only some newborn CHT is genetic
• Cystic fibrosis
• Deafness: some newborn deafness is genetic
• Phenylketonuria (PKU)
• Sickle cell disorders
• Medium chain acyl-CoA dehydrogenase deficiency (MCADD)

This area of healthcare is developing quickly and it will be important for you to use these sources to gain relevant information when you need it and to keep up to date, building on the basic knowledge in this chapter. As in all areas of practice, however, it is important to understand the limitations of your own knowledge and, where necessary, refer parents to specialist practitioners or colleagues with more advanced knowledge of genetics.

ScotGEN - www.scotgen.org.uk
Professional Education for Genetic Assessment and Screening (Pegasus) - www.pegasus.nhs.uk
National Genetics Education and Development Centre - www.geneticseducation.nhs.uk
Genes, chromosomes and DNA

This chapter will help you become familiar with a number of important key words such as genes, deoxyribonucleic acid (DNA), chromosomes and karyotype.

Our genes are the set of chemical instructions that direct the development, growth and function of every cell of our body, from conception to death. We inherit our genes from our parents, therefore we can think of a gene as a unit of inheritance which can be copied and passed on to the next generation. Each gene is made of a section (or segment) of DNA. This is a long thread-like molecule that makes up structures in our cells called chromosomes (Figure 1).

Our 25,000 genes, each of which contains information or codes for specific characteristics or functions, are organised in sequence along our 23 pairs of chromosomes (like beads on a necklace) that are present in the nucleus (the control centre) of most human cells.

Cytogeneticists and molecular geneticists are clinical scientists who specialise in looking at cells. Cytogeneticists can prepare and analyse the set of chromosomes present in human cells, while molecular geneticists concentrate on examining DNA. Depending on the indication for the diagnostic test, liquor, placenta or cord blood can be sent to a cytogenetic laboratory, a molecular...
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Understanding Genetics

genetic laboratory or both. The 23 pairs of human chromosomes can be displayed as a microscopic preparation then viewed using a microscope, enabling an experienced cytogeneticist to distinguish the chromosomes from each other.

A microscopic preparation and description of the chromosome structure of an individual is called a karyotype. It includes the number of chromosomes and any variation from the normal pattern.

Figures 2 and 3 show karyotypes of a female and a male with the usual 23 pairs of chromosomes. If you look at them closely you will see that there are both similarities and differences between them.

Figure 2.
A female karyotype with the usual 23 pairs of chromosomes

Normal Female 46, XX

Figure 3.
A male karyotype with the usual 23 pairs of chromosomes

Normal Male 46, XY
The chromosomes in each matched pair within the karyotypes are similar in shape, size and patterning. Individual chromosomes are similar in all humans, even if some genes on them give the cells of the body very different instructions from one human to another.

We inherit one chromosome in every pair from each parent. The only pair that is not similar is the pair of sex chromosomes in the male karyotype. This is the difference between male and female karyotypes.

The 22 pairs of chromosomes are called autosomes and the X and Y chromosomes (the 23rd pair) that are responsible between them for determining the sex of an individual are known as sex chromosomes.

Because women have two X chromosomes, they always pass on one of these to their children. However, men have one X and one Y chromosome, so the sex of a baby depends on whether it inherits its father’s X or Y chromosome.

Figure 4 shows a karyotype of a female with Down’s syndrome. You can see that the karyotype is female because there are two X chromosomes. The difference between this and a normal karyotype is the extra chromosome 21 (trisomy chromosome 21). There are 47 chromosomes present instead of the normal 46.

More genes, chromosomes and DNA

Each chromosome contains a molecule of deoxyribonucleic acid (DNA), with hundreds to thousands of genes arranged along its length. For example, chromosome 21, the smallest human chromosome, has more than 300 genes. Genes usually code for proteins, which perform a wide range of specialised functions. For example they might fight infections, turn other genes on or off, form structures such as heart muscle, haemoglobin (the oxygen carrier in red blood cells), and transmit messages between cells.

The Australian Centre for Genetics Education (http://www.genetics.com.au) explains the relationship between genes and chromosomes in a clear, simple way:

Your genome, or ‘Book of Life’, can be thought of as two volumes, one inherited from your mother and one from your father. Each book has 23 chapters (the chromosomes), containing many pages (the genes). The words on each page make up information and instructions (the message each specific gene gives to cells).

The genetic code

The DNA molecule contains four building blocks or bases:
- Adenine (A)
- Thymine (T)
- Guanine (G)
- Cytosine (C)

The genetic code is based on groups of three bases (eg, ATT, CGT), which each specify an amino acid that provides the building block for a protein. Errors in the code can result in the wrong amino acid being put in, or the protein chain being shortened or altered. This can have a marked effect on the structure of the protein and the way it works.

An individual’s physical characteristics and physiology, their predisposition to health and disease all depend on the functions of their inherited genes within their body cells and responses to the environment. The next section looks at the way genes contribute to health and ill-health more generally.
Part One
Understanding Genetics

Genetics, health and disease

Variation

All of us, except those who are identical twins, have different genetic make-ups and we all experience differences in our environment and upbringing. This combination of differences in genetic make-up and environmental exposure, ‘nature’ and ‘nurture’, results in the differences among us and makes each one of us unique. Genetic variation and diseases result from alterations in the DNA sequence, either at a gene or chromosome level. When chromosomes and their genes are copied to form new cells – either sex cells (eggs/ova or sperm) or body cells – an error may occur. Most errors are repaired but if they are not errors in the copying or a mistake in the way the chromosomes are passed on to the new cells will result in a gene or chromosome alteration. If this alteration is present in the sex cells, it can be passed on to a baby and may cause a disease or genetic disorder.

Understanding different types of disease-causing mutations will support your understanding of how these alterations may be passed on from one generation to the next or may arise spontaneously without any previous family history of the condition. Below are brief descriptions of the ways in which genes can be involved in diseases: single-gene disorders, chromosomal anomalies (abnormalities) and multifactorial diseases.

‘Mutation’ is the technical term for an alteration in the DNA sequence at gene or chromosome level. However, this term is usually avoided in discussions with parents in favour of neutral terms such as ‘alteration’, ‘anomaly’ or ‘change’. Other negative terms such as ‘abnormal’ and ‘defective’ and ‘faulty’ are also usually avoided.

Genes and disease

Single-gene disorders

These include cystic fibrosis, sickle cell disorders, thalassaemias, Duchenne muscular dystrophy and Huntington’s disease.

Inherited single-gene disorders are associated with mutations in a single gene, which can cause the protein product of that gene to be altered or missing. This then results in a disorder or disease being expressed – the gene produces a protein that can result in a disorder. These disorders are inherited in distinct patterns that are understood and used by specialists involved in clinical genetics to advise people about chance of genetic disease to themselves or their family. The patterns of inheritance vary according to whether autosomes or sex chromosomes (the X or the Y chromosome) are involved and whether the alteration needs to be present on both copies, or only on one copy, of the gene to be expressed. We discuss this further in the section on patterns of inheritance.

Chromosome anomalies

Chromosome alterations are known as chromosome anomalies. The most common condition we encounter in which a chromosomal anomaly causes learning disability is Down’s syndrome. You saw a karyotype for a female with Down’s syndrome in Figure 4 in the section on chromosomes (page x). The person with Down’s syndrome had a different, or anomalous, karyotype in which there were three copies of chromosome 21.

Turner syndrome is another example of a chromosome anomaly, where a female has only a single X chromosome instead of the usual two. In Turner syndrome there are again characteristic clinical features, the two most common problems being short stature and ovarian failure. There is more on Turner syndrome on the Turner Syndrome Support Society website: http://www.tss.org.uk

Chromosome anomalies occur when whole chromosomes or large segments of chromosomes are duplicated, missing or altered. They happen when the chromosomes are copied then distributed between newly forming sex cells. When a sex cell with a chromosome anomaly from one parent combines during fertilisation with a sex cell from the other parent, the fertilised egg carrying a chromosome anomaly has the potential to develop into a new individual. Most chromosome anomalies are lethal to the fetus so a miscarriage results, but some are less severe and can be compatible with a full-term pregnancy, though the baby may have disabilities.
A whole extra chromosome is called **trisomy**. A missing chromosome is known as **monosomy**.

### Multifactorial disorders

Although some alterations in our genetic make-up are harmful and may cause disease, others have no noticeable effects or may even be useful. Everyone has up to 100 genetic alterations, which form the genetic variation (differences) among people. These natural differences in genetic make-up between members of a population are sometimes known as natural genetic variation and are essential to enable any population to survive and evolve when faced with changes in their environment.

This normal variation among people may also cause variation in susceptibility to multifactorial disorders such as spina bifida, diabetes or coronary heart disease. An increased chance of these diseases results from interactions between several different genes, which may be normal gene variants, and environmental factors (such as diet, smoking or exposure to radiation). Too many negative factors, both genetic and environmental, can tip the balance towards expression of the disease. These diseases may tend to recur in families because there is a greater chance that related people will share the same sets of genes (normal gene variants) but there is no clear pattern of inheritance (in contrast to the single-gene disorders).

### Inheritance Patterns

Single-gene disorders (monogenic disorders) such as cystic fibrosis and Huntington’s disease are caused by a mutation in one gene, which can be passed from one generation to the next.

They have simple patterns of inheritance, which make it possible to predict the chance of a child developing a particular disorder if the genotype of both parents is known with respect to that disorder – if we know which versions of the genes each parent possesses. However, even with the same mutation, not everyone in a family will have identical medical problems or be equally affected. This variable expression means that the altered gene expresses itself in different ways in different people.
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Understanding Genetics

The patterns of inheritance were first described by Gregor Mendel, a 19th century monk. Hence single-gene disorders are also known as Mendelian disorders.

Our genetic make-up, our **genotype**, describes all the genes we have inherited, whether they are expressed as our characteristics or not, whereas our **phenotype** describes how our genes are expressed – our physical appearance, physiological molecular or biological traits. All phenotypes are the result of an interaction between our inherited genotype and the environment.

To understand inheritance patterns you need to concentrate on how we inherit our genes and how these genes are expressed. We inherit one copy of each of our genes from our mother and father. They may both give us the same version of the gene, or they may give us different versions. For example, one may give us the gene for blue eyes and the other the gene for brown eyes. The gene that is expressed, which determines our eye colour, is the dominant gene. These different versions of the same gene are called alleles.

There are three main types of inheritance patterns for single-gene disorders:

- **Autosomal dominant inheritance**
- **Autosomal recessive inheritance**
- **Sex-linked recessive inheritance**.

### Autosomal dominant inheritance

The presence of a dominant gene (dominant allele) determines the characteristic that is expressed. An autosomal dominant disease is developed when an individual inherits the faulty dominant gene (dominant allele) from one parent (Figure 6).

**Example**

Huntington’s disease is an autosomal dominant disorder associated with the presence of an autosomal dominant allele. For the disease to develop in an individual he or she needs only one copy of the altered allele. Because the condition is associated with nonsex chromosomes, males and females are equally likely to be affected.

An affected person usually has an affected parent and there may be other family members affected such as grandparents, siblings, uncles, aunts or cousins. An individual with a parent who is affected has a 50% chance (one in two) of having inherited the altered allele and so being affected with the disease.

Sometimes a child is born with a dominant disorder although neither parent is affected. This is frequently the case with achondroplasia. This is because a copying error (mutation) occurred when the egg or sperm was being made.

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**Figure 5. Autosomal dominant inheritance (Mendelian pattern of inheritance)**

- **Affected Parent**
- **Unaffected Parent**
- **Affected child**
- **Unaffected child**

- **Altered gene**
- **Normal gene**
In the past, carriers of the haemoglobin disorders such as sickle cell disorder were said to have sickle cell trait. The term ‘carrier’ is now used, in common with other inherited genetic disorders.

Example
Cystic fibrosis (CF) is an autosomal recessive disorder. For a child to develop the disease it needs to inherit altered copies of the gene from both parents.

Both parents must then be carriers of the altered version of the gene (the disease causing allele). Each time conception occurs between two carrier parents there are four possible outcomes because in each egg or sperm there is an equal chance (50% or one in two) that there will be a normal or an altered copy of the gene.

This means there is:
- A 25% (one in four) chance that the child has received two normal copies of the gene and is unaffected and not a carrier
- A 50% (two in four) chance that the child has received one normal copy of the gene and one altered copy and is an unaffected carrier.
- A 25% (one in four) chance that the child has received two altered copies of the gene and is affected.
- Males and females are equally likely to be affected.

For further information on CF, see the Cystic Fibrosis Trust website: http://www.cftrust.org.uk

The occurrence of an autosomal recessive disorder relies on the chance that two individuals who are both carriers (although they may be unaware of this) produce offspring. All of us carry recessive genes that might affect our children if our partner also carried the same recessive genes. Because people in families are more genetically similar to each other, these autosomal recessive conditions occur more frequently when parents are related to each other (the relationship here is said to be consanguineous). For similar reasons certain diseases such as Tay-Sachs disease occur more commonly in particular ethnic groups.

Tay-Sachs disease is a rare disorder in which there is a progressive degeneration of all brain functions. It appears in early childhood and usually leads to death in early childhood. It is common among Ashkenazi Jews – a group of Eastern

Some other examples of autosomal dominant conditions include:
- Marfan syndrome
- Neurofibromatosis
- Familial adenomatous polyposis (a form of colon cancer)

Autosomal recessive inheritance

For an autosomal recessive disease to be present in an individual both copies of the gene need to be altered (Figure 7). An individual with one normal and one altered version of the gene (or allele) will not develop the disease but will be an unaffected carrier. If one parent is a carrier and the other is not, there is a 50% chance that their child will be an unaffected carrier.

For further information on CF, see the Cystic Fibrosis Trust website: http://www.cftrust.org.uk
European Jews – in whom about one in 30 people carry the gene. You can find out more about Tay-Sachs disease on the Children Living with Inherited Metabolic Diseases website: http://www.climb.org.uk

**Sex-linked (X-linked) inheritance patterns**

Females inherit two X chromosomes while males inherit only one. The X chromosome carries many important genes that have nothing to do with determining sex. If a female has an altered gene for a genetic disorder on one of her X chromosomes, she is likely to have an unaltered gene for that disorder on the other X chromosome and will therefore be an unaffected carrier, or in some cases mildly affected. Since males have only one X chromosome the altered gene will not be compensated for by another copy of the gene and a male will therefore be affected.

*Example*

Duchenne muscular dystrophy (DMD) is a sex-linked genetic disorder caused by an X-linked recessive gene alteration. This means the gene associated with this disease is located on the X chromosome. Males (XY) are predominately affected because the recessive DMD allele is present on the X chromosome. If a male inherits an altered copy of the gene, he will have no unaltered copy to compensate for it and so will have DMD.

Sons of carrier females have a 50% chance (one in two) of inheriting the DMD allele and developing the disorder. Daughters of carrier females have a 50% chance (one in two) of inheriting the DMD gene but will usually show no signs of the disease (see fig 7).

*Duchenne muscular dystrophy is a condition in which there is progressive muscular weakness because muscle cells break down due to a defect in dystrophin an important protein in muscle fibres. It affects only boys (with extremely rare exceptions), who first show difficulty in walking between ages one and three. The condition is progressive with a shortened life expectancy; most die by the late teens or early twenties.*

You can find out more about Duchenne muscular dystrophy on the Muscular Dystrophy Campaign website: http://www.musculardystrophy.org

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Figure 7. Sex-linked inheritance where the mother is a carrier and the father is unaffected

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Other examples of sex-linked conditions are:

- Haemophilia
- Hunter syndrome
- Colour blindness
- Fragile X syndrome.
The newborn blood spot test (the heel prick which tests for PKU, CHT, sickle cell disorders, cystic fibrosis and MCADD) aims to identify genetic disease at an early stage so that treatment can be started early. In the case of PKU this means a diet low in phenylalanine, which can reduce the neurological damage associated with PKU. Some diagnostic genetic tests for recessive conditions such as sickle cell disorder can identify the carrier status in the mother during pregnancy. If this test is positive the father can also be offered a test. If both parents are carriers there is a chance that the fetus will be affected or an unaffected carrier. Diagnosis could then be offered via amniocentesis or chorionic villus testing, depending on gestation.

In the pregnancy period identifying genetic disease can allow parents and health services to be prepared for the birth of a baby who may have specific health problems, and, if the condition is likely to lead to severe problems, it may give them the opportunity to consider whether or not they want to continue with the pregnancy. These are all examples of disease prevention in its wider sense – that is both the prevention of disease and the reduction of its severity.

The purpose of genetic screening is to support parents to improve the health of their families and enable them to make informed choices about a pregnancy where the baby is likely to have a serious disease or condition. It is not to prevent the birth of babies with the condition in question.
Ethical, legal and social dimensions of genetic testing

There are many implications of genetic testing that go beyond the usual areas of concern in clinical practice: those focusing on promoting health, preventing, diagnosing and treating disease, and providing holistic care for families. These implications are often described as the ethical, legal and social issues in genetics (sometimes abbreviated to ELSI issues).

You can find out more about them on the following websites:

- Public Health Genetics Unit: http://www.phgu.org.uk
- National Human Genome Research Institute: http://www.genome.gov

It is important to be aware of these implications and complexities so that you can be prepared for them as you discuss tests with patients. However, you must recognise the limits to your practice. Clinical geneticists, genetic counsellors and some other specialists (e.g., haemoglobin disorder counsellors) are specially trained and experienced in dealing with these areas and will often do so as members of a multidisciplinary team. You should therefore make appropriate referrals to those experienced in answering difficult questions and managing complex genetic issues.

Identifying a fetus with a serious genetic condition means that parents have to make a choice about whether to continue with the pregnancy. For individuals and society this is a serious ethical and moral decision and one that can have profound consequences for parents and families. For example:
Improved understanding of genetics promises a future of precise, individualised medical treatments. Pharmacogenetics, the study of how different people respond to drugs due to their genetic make-up, could lead to customised drug treatments. The hope is for targeted and effective treatment with few side effects.

Gene therapy is a technique based on the principle of replacing or modifying a faulty gene with a normal functioning gene in the target tissues to reduce or prevent the expression of a disease. Although it remains an experimental treatment ongoing research may eventually provide cures for genetic diseases such as cystic fibrosis.

Stem cells are the precursors of each of our highly specialised body cells. Stem cell research and an understanding of the genetics of these cells could lead to the ability to replace and repair any damaged tissues and organs in our bodies. So in the future 'new' functioning individualised tissues and organs may be available to treat a range of diseases eg, insulin-producing cells for people with diabetes, skeletal muscle cells for muscular dystrophy, neural (nerve) cells for neurodegenerative diseases and spinal cord injury.

Future developments

The Human Genome Project began in 1989 and succeeded in producing a virtually complete sequence of our human genetic make-up in 2003. This detailed and comprehensive analysis has paved the way to the next stages of interpretation; identification of all of our estimated 25,000 genes and their functions. These leaps in genetic knowledge will affect all areas of medicine in the near future.

You can find out more about the Human Genome Project at the Wellcome Trust Sanger Institute (http://www.sanger.ac.uk) and the Wellcome Trust (http://www.wellcome.ac.uk).

The scientific understanding and knowledge gained from the Human Genome Project has implications for future prevention, diagnosis and treatment of disease. Developments in our knowledge of genetics will improve our understanding of health and disease, facilitating earlier diagnoses, providing opportunities for timely and targeted interventions which could be combined with more effective and better treatments.
Part One
Understanding Genetics

Conclusion

This chapter should have helped you to develop your understanding of the basics of genetics as it applies to your practice. This should help you when working with women, their partners and families and helping them to make informed choices in relation to pregnancy and newborn screening for genetic conditions. Where situations are outside your knowledge it is vital to refer parents on to specialist genetics services.

You should now be able to:

1. Understand the basics of human genetics.

2. Discuss the role of genetic factors in health and disease.

3. Explain the mode of inheritance in single-gene (Mendelian) disorders (dominant, recessive and sex-linked recessive) and provide examples of specific conditions.

4. Acknowledge the role of specialist genetic services in health services and how genetic tests can have implications for prevention of disease and promotion of health.

5. Understand how genetic services and pregnancy diagnostic services relate to each other.

6. Discuss the developments in genetics for future healthcare.
Part One
Informed Choice in Pregnancy and Newborn Screening

**Informed choice in everyday life**

There are many areas in life where it is important to be able to make an informed choice in order to limit the occurrence of potentially harmful and/or costly consequences.

Armed with this information, different people will make different choices based on the needs and preferences of their group. For example, when planning a journey, cost may be a priority for some, while comfort or journey time may be more important to others.

In pregnancy and newborn screening it is the responsibility of health professionals involved at all stages of the screening process to ensure women, their partners and families are enabled to make informed choices. In order to do this, professionals need to understand the concept of informed choice, the processes required to achieve it and the issues that may affect individuals’ needs and their abilities to make informed choices.

Before discussing informed choice in relation to pregnancy and newborn screening, you may find it helpful to explore informed choice as it relates to healthcare more generally.

**Informed choice in healthcare**

Screening can be an important tool in providing effective care and increases the reproductive choices available to women; however, findings can have potentially serious medical, social or financial consequences for the individual, their child and their relatives.

It is essential that anyone being offered screening for themselves or their child has access to clear information presented in a way that enables them to make a properly informed decision. They need time to consider information and must be supported both practically and emotionally by staff during this process.

Prior to deciding whether to accept or decline screening they should have a clear explanation of:

- Whether possible to opt to have a diagnostic test rather than screening;
- The purpose of the particular screening test and what the test involves;
- The likelihood of positive/negative findings and the possibility of receiving a false positive/negative result;
- The limitations and risks attached to the screening process;
- Any significant medical, social or financial implications of screening for the particular condition or predisposition;
- Follow up plans, including how and when results will be obtained, availability of counselling and support services.

Consent is only valid if these key elements are complied with:

- The consent is given voluntarily, with no pressure or coercion.
- Appropriate information has been received in the most appropriate format and the consequences of screening and any results are understood.
- The person has capacity.

**Definitions of informed choice**

A number of definitions have been proposed for informed choice. The four we have included here all refer to maternity services or pregnancy or newborn screening:

‘Informed choice in maternity services:

- Is a partnership between woman and a health professional?
- Involves exchanging information, communicating with and listening to each other
- May take time for women to make decisions
- Involves presenting and considering more than one option
- Takes into account evidence, chances and benefits
- Respects the woman’s autonomy
- ‘Ideally results in a decision being made and owned by the woman who is then supported by the health professional.’ (MIDIRS, 2004)

‘Informed choice has been described as an expression of respect for the autonomy of the client and respect for her right to self-determination regarding reproductive choices.

Informed choice requires complete, objective information provided in the context of a non-authoritarian, collaborative relationship and the
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active involvement of the client.’
(Harding, 2000)

‘An informed choice is one that is based on
relevant knowledge, consistent with the decision-
maker’s values and behaviourally implemented.’
(Dormandy et al, 2002)

Elements of informed choice in newborn screening

• ‘Decisions are made voluntarily by parents:
  • Who have considered and understood
    information about screening, including the
    benefits and chances
  • In line with parents’ own values, beliefs, wishes
    or priorities
  • To accept or decline screening for all or some
    of the conditions
  • To accept or decline tests that identifies carrier
    status
  • To accept or decline follow-up screening or
    diagnostic tests
  • To accept or decline invitations to participate
    in research relating to the newborn blood spot
    screening programme. (Hargreaves et al,
    2005)

You may consider that informed choice has
potential disadvantages because, in a resource-
limited service, it could take more time to enable
women and families to make decisions about
offers of screening. You may also think it could
cause difficulties for some women and families
in making often complex decisions. However,
understanding the process through which informed
choice is achieved, the conditions for which
screening is offered and the issues women and
families must consider can help you to support
them through this process in a systematic way,
making the best use of your time and enabling you
to understand when referral to professionals with
more specialist knowledge is required.

Pregnancy screening

Although most mothers and babies are healthy,
screening tests may indicate an increased risk of
serious disorders in a small proportion of them.
Screening can therefore lead to further decisions
about whether to have invasive diagnostic tests –
which have associated risks – and even whether to
continue with a pregnancy. Such situations involve
complex choices for women and their families
and it is vital that they receive the information and
support they need to ensure they can make an
informed choice that is right for them.

Other pregnancy screening tests are simple, carry
little or no associated risk and will benefit the
woman and/or her baby. For example, tests for
infectious diseases in pregnancy may involve a
straightforward decision, as treatment is available
which can have health benefits for both the woman
and the baby. However simple the tests may
seem, it is still important to ensure women can
make fully informed choices about whether or not
to accept the tests – they should not be treated
simply as a routine procedure.

In the case of HIV, hepatitis B or syphilis, some
women find the decision whether or not to accept
a test more complex for a range of practical and
emotional reasons – even though diagnosis would
give them access to treatment that would benefit
both them and their babies. For example, if a
woman’s partner is present, she may feel that
to accept the tests would imply that either she
or her partner had engaged in behaviours that
could put them at risk of contracting the diseases.
Alternatively, she may feel that the fact that she
is being offered the tests implies a judgement is
being made about her.

Attitudes to HIV and AIDS are not as negative
as they were in the early years after they were
identified as a serious health issue. However,
there is still a stigma attached to them in the
eyes of some people in both developed and
developing countries (Brown et al, 2003). This may
be because they see HIV as being the result of
‘immoral’ or ill advised behaviour, or because they
do not understand how the virus is transmitted
(Letamo, 2003).

In addition, some women may believe that simply
having an HIV test could affect their ability to
obtain life or health insurance. This belief is based
Newborn screening

Newborn tests could be considered relatively simple in most cases, as they carry little or no risk to the baby, while if one of the conditions in question is diagnosed, treatment is available.

In addition, early intervention is important in the case of phenylketonuria (PKU) and congenital hypothyroidism (CHT) to reduce the chance of long term disability. However, parents should still be enabled to make informed choices about whether to accept or decline the screening tests offered for their babies. In order to do this they need the same level of information as they receive about offers of pregnancy screening.

Parents who accept blood spot screening should also be told that after testing, blood spots are stored for an initial period of 12 months in the laboratory, in case any of the screening tests need to be repeated to check a particular result. The stored blood spot specimen can also be used to test for some other disorders which are not part of the standard screening programme. This may be useful if a child becomes ill and the doctor requests further tests, but this would always be discussed with the child’s parents first. Left over blood spot specimens can also be used anonymously for other laboratory purposes such as research.

Health professionals discussing the offer of HIV screening must therefore take care to ensure the woman has been given, and understands, all the relevant information about transmission of HIV and the benefits of antiretroviral therapy if she is found to be infected. They should also emphasise that the test is offered to all pregnant women, and that the offer does not suggest she may be in a higher-chance group. She should also be reassured that her ability to obtain life insurance will not be prejudiced if she is not infected with HIV.
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as comparing different screening methods and developing new tests. Occasionally it is necessary to use identifiable specimens in which case the parents’ permission would always be sought. If the parents do not want the stored blood spot card to be used for further research the health professionals collecting the blood sample should indicate ‘No Research’ on the blood spot card.

Whatever pregnancy and newborn screening they are offered, it is vital that women and families understand what the test is for and what it involves. They must also understand that they have a choice whether or not to accept it. There are a variety of links providing further information and advice:

- Pregnancy Results and Choices (ARC: http://www.arc-uk.org) provides professional education for practitioners involved in screening.
- MIDIRS informed Choice leaflets: www.midirs.org

In order to help women and their partners to make informed choices, health professionals need to understand what informed choice constitutes.

**Ensuring informed choice in screening: essential information**
- Testing is optional – at any time
- What is being screened for and the implications
- Timing of various tests
- Results: when and how they will be delivered
- What the possible results mean
- Detection rates of various tests
- Options after a result indicating increased risk of an affected baby
- Reassurance that most pregnancies and babies are normal and healthy.

Then check parents’ understanding, give a telephone number in case they have further questions, and written information on what they have been told.

**Giving parents’ time**

Unlike many major decisions in life, those related to pregnancy and newborn screening must often be taken quickly:

- Pregnancy tests must be carried out at particular stages in a woman’s pregnancy, often because there is an optimum window of time for detection of markers for particular conditions in the fetus, placenta or mother.

This information is necessary to enable parents to make decisions such as whether to accept diagnostic tests or continue with the pregnancy, while decisions such as whether or not to terminate also need to be taken quickly.

- Newborn screening tests are offered within days or weeks of a baby’s birth. Some tests, such as those for PKU and CHT rely on early identification and treatment of affected babies in order to prevent severe learning disabilities. If parents decline these tests and then change their minds, the tests will still be available, but the benefits for an affected child will not be so great. Other tests, such as physical examination, can enable early interventions that treat conditions in affected babies more easily than in older children.

Although parents often have to make these important decisions in a relatively short time, it is vital that they are enabled to make informed choices. If they are rushed into making a decision without having received and understood all the relevant information and considered the implications of each possible choice – and received any support they need, the emotional effects can be extremely long-lasting and detrimental.

Informing for choice is part of a continuum in pregnancy and newborn screening in which women and families are offered a range of tests, possibly with further choices after receiving test results. They then make their choice about how to proceed, and finally they give or withhold their consent to any procedures. In order to ensure the choices are informed, health professionals go through a process that enables women and
families to understand the tests and conditions in question, and their implications for both themselves and their baby. This can involve any, or all, of informed decision making, shared decision-making and supported decision-making in a single consultation, depending on the individual needs of the woman, her partner and/or her family.

Informed choice and informed consent

‘Informed consent is an ongoing agreement by a person to receive treatment, undergo procedures or participate in research, after chances, benefits and alternatives have been adequately explained’ (RCN, 2004).

When screening or diagnostic tests have been offered to women and families, provided they have been given all the information and support they need – and time to consider this, they will be able to make an informed choice about whether to accept or decline the offer. If they choose to accept it, they can confirm their decision to the health professionals providing their care by giving their informed consent for themselves or their baby to undergo the procedure. If they choose to decline the offer, this decision is similarly informed.

Informed consent is an important legal and ethical principle in healthcare, reflecting patients’ rights to determine what happens to their own bodies. The quote below describes informed consent in more detail.

‘Informed consent is the process by which a fully informed patient can participate in choices about her health care. It originates from the legal and ethical right the patient has to direct what happens to her body and from the ethical duty of the physician to involve the patient in her healthcare.’

What are the elements of fully informed consent? ‘The most important goal of informed consent is that the patient has an opportunity to be an informed participant in her healthcare decisions. It is generally accepted that complete informed consent includes a discussion of the following elements:

• The nature of the decision/procedure
• Reasonable alternatives to the proposed intervention
• The relevant risks, benefits, and uncertainties related to each alternative
• Assessment of patient understanding
• The acceptance of the intervention by the patient.’ (Edwards, 1998)

Legal issues in informed consent

As in all areas of healthcare, obtaining informed consent is a legal requirement before any pregnancy or newborn screening test is undertaken. This is vital to protect both the people offered screening and the health service.

A woman can only give informed consent to a test on herself or her baby if she has been able to make an informed choice about the options available. According to the Department of Health (2001b) she must:

• Be competent to take the particular decision
• Have received sufficient information to take it
• Not be acting under duress.

If a woman accepts or declines a test but does so without fully understanding its implications or because she feels she is expected to do so, she may make a decision that is not suitable for her.

This may cause unnecessary harm to herself or her baby and put her healthcare provider at risk of litigation. Informed consent is therefore important for a number of reasons including:

• To ensure the best possible outcomes for women and their babies
• To protect healthcare providers from litigation
• To protect health professionals from complaints to their employers or professional bodies.

If a test is undertaken without a woman’s consent, she can take legal action for assault/battery or negligence. It is essential for health professionals to document all tests offered in women’s records, the information and support they are given and that they have given informed consent if they choose to accept any of the tests, or made an informed refusal. Without detailed records of this information, made at the time of the consultation or as soon as possible after, healthcare providers have no evidence that the care they offered was appropriate and sufficient. This can be important in cases where adverse outcomes lead to legal actions against the provider.
In some cases a woman may give implied, rather than expressed, consent to a procedure. For example, she may extend her arm to have her blood pressure tested or prepare for an ultrasound scan by lying back and uncovering her abdomen. Again, however, it is still important for the health professional to ensure that she has made an informed choice to consent to or decline the test. Implied consent is not necessarily informed consent.

Written consent

In some situations, such as when they are offered hearing screening for their babies, women are asked to sign forms stating that they consent to or decline particular tests.

However, while a signature on a form is evidence that a woman has given consent, it does not prove that the consent is valid. If she is rushed into signing a form without the information she needs to make an informed choice, it will not be valid. Similarly, even if she signs a consent form, she may, if she wishes, withdraw her consent at any time if she changes her mind – the consent form is not a binding contract. The important point to remember when seeking written consent is that the same process must be gone through to ensure the woman is making an informed choice.

Although properly informed written consent can be useful to document a woman’s decision, in many cases it is not necessary, particularly in routine or low-risk procedures.

However, it is good practice to seek written consent if:

- The procedure is complex or involves significant chances of an adverse outcome
- The procedure involves general or regional anaesthesia or sedation
- Providing clinical care is not the primary purpose of the procedure
- There may be significant consequences for the woman’s employment, social or personal life
- The procedure is part of a research programme
- You have reason to believe the consent may be disputed later.

Bridson et al (2003) and the General Medical Council (1998) emphasise that if they are to obtain truly informed consent health professionals must consider patients’ own needs and priorities. This involves making oneself aware of their past experiences, beliefs, cultural norms and any other factors that may have a bearing on the their attitudes to screening, the information they need in order to make a decision and ensuring these needs are met. It is important not to make assumptions about what these needs and priorities might be.
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Written consent can also be helpful to ensure health professionals are clear about what the woman is consenting to when more than one test can be taken. For example, in the case of pregnancy blood tests, it is important to ensure the woman knows exactly what conditions she will be tested for, and that she knows she can accept or decline any or all of them. Similarly, it may be useful to gain written consent confirming what is to be tested for in pregnancy diagnostic tests.

A woman may accept a CVS or amniocentesis to look for a particular condition such as Down’s syndrome or sickle cell disorder without realising that the test can also pick up other conditions. In the case of newborn blood spot screening the UK Newborn Screening Programme Centre recommends that parents’ decisions about whether to accept or decline screening for their baby should be recorded in the maternity notes, along with a record of the discussion that took place to enable them to make their decision and in Scotland, parents are asked to give written consent.

## Ability to give consent

Another area in which there are legal implications relating to informed choice in pregnancy and newborn screening is where the woman may not be competent to make decisions for herself, either because she is too young or due to mental incapacity because of mental health problems or learning disabilities. In general, those aged 16 years and older are considered capable of giving their own consent (unless their intellect does not permit this). However, the Age of Legal Capacity (Scotland) Act 1991 states that ‘a person under the age of 16 years shall have legal capacity to consent on his own behalf to any surgical, medical or dental procedure or treatment where, in the opinion of a qualified medical practitioner attending him, he is capable of understanding the nature and possible consequences of the procedure or treatment’ and therefore to have the capacity to give informed consent, or be involved in decisions about their care.

In the case of pregnancy and newborn screening offered to mothers under 16 years of age, health professionals need to assess the individual mother’s capacity, depending on her maturity and intelligence, in order to determine whether or not she is competent to make informed choices for herself (RCN, 2004). The same is true for women with mental health problems or learning disabilities – health professionals need to assess the individual woman’s capacity to understand the choices being offered and to make rational decisions for herself. In these cases, parents, carers or partners cannot override the woman’s consent if she is considered to be capable of making decisions for herself.

See the Scottish Good Practice Guidelines for Supporting Parents with Learning Disabilities for further information and advice.

My Pregnancy, My Choice, published by CHANGE is an easy read handbook for parents with learning disabilities and is available free of charge from Health Scotland.

## Parental Responsibility

Remember that not all parents have parental responsibility.

Legally, both parents will have parental responsibility if they were married at the time of the child’s conception or birth, or at some time after the child’s birth. Neither parent loses parental responsibility on divorce.

If the parents have never been married, only the mother automatically has parental responsibility, but the father may acquire that status by order or agreement. There are current proposals to change the law in this area.

Parents who do not have parental responsibility nonetheless play an essential role in determining best interests and may have a right, under the Human Rights Act, to participate in the decision-making process.

In some circumstances, people other than parents may acquire parental responsibility, for example by the appointment of a guardian or on the order of a Court.

If there is any doubt about whether the person giving consent is legally entitled to do so, legal advice should be sought.
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Ethical issues in informed choice

Informed choice and informed consent do not only have legal implications – they also have important ethical implications. At the most general level ethical perspectives are important because, as the Committee on Bioethics (1995) points out, informed consent reminds health professionals to respect patients ‘by fully and accurately providing information relevant to exercising their decision-making rights’. Respect is the core ethical principle in all aspects of healthcare, and in pregnancy and newborn screening it involves taking into account each woman’s individual needs, considering factors such as:

- Ethnicity
- Gender
- Disability
- Religious beliefs
- Culture
- Language
- Level of understanding

In order to achieve this, health professionals need to act professionally, be sensitive to individual needs and to avoid making assumptions when discussing women’s choices in relation to screening, and gaining their informed consent for the choices they make. Ethical dilemmas can be extremely complex and difficult to handle. While knowledge of the law and of ethical theories can offer you some help in working through these difficult situations, they cannot provide solutions to all dilemmas. The requirements of your own professional code of conduct can offer guidance on how to work through dilemmas in a professional and sensitive way. Each individual situation must be dealt with on the basis of the specific circumstances of those involved. Whether or not you are able to offer the help parents need, will depend on your own professional skills and knowledge. The Nursing and Midwifery Council (NMC) state that:

- ‘you must treat people as individuals and respect their dignity’
- ‘you must act as an advocate for those in your care, helping them to access relevant health and social care, information and support’
- ‘you must listen to the people in your care and respect their concerns and preferences’
- You must make arrangements to meet peoples language and communication needs
- You must ensure that you gain consent before you begin any treatment or care
- You must uphold people’s rights to be fully involved in decisions about their care
- You must be aware of the legislation regarding mental capacity, ensuring that people who lack capacity remain at the centre of decision making and are fully safeguarded

The code: Standards of conduct, performance and ethics for nurses and midwives. (2008) NMC
Enabling decision-making

Although Coulter and Magee (2003) found that patients want greater involvement in decision-making about their healthcare, this does not mean all patients want to make all decisions for themselves or that they all want the same level of involvement. This applies equally to pregnant women and new mothers and their partners and families in relation to pregnancy and newborn screening, which is why maternity services must enable them to be active participants in planning their care. While health professionals working in pregnancy and newborn care are increasingly expected to encourage their clients to make informed choices about screening, this may not suit all those in your care. You may find that most are confident about making decisions for their selves once they have been given the information they need to make an informed choice, but some are likely to ask for more guidance. This may range from simply asking ‘What do most people do in this situation?’ through to saying ‘We can’t make that decision – you decide for us.’

When women and their partners ask what most people do in their situation it may be relatively easy to deal with by explaining that everyone is different, and what is right for one person may be wrong for another. If they ask for statistics or for your experience, you can emphasise that what most people do is not necessarily the right thing for them, and that they should still think about their own circumstances. Again, it is vital that the information you give is balanced to enable women and their partners to make informed choices.

However, if a woman and her partner ask you to make a decision for them, the situation is more difficult to deal with. Even if you have a strong opinion about what they should do, in your role as a health professional it is not your decision to make. Your professional role should be to help your clients to work through the issues as they apply to them, while ensuring they remain in control of the decision. You can, if they insist, offer your professional opinion but stress that the decision must be theirs.

Conclusion

Ensuring women are able to make informed choices about pregnancy and newborn screening can be a complex and demanding task. Although it is not possible to provide answers to every situation you are likely to encounter, we hope this chapter has helped to increase your awareness of some of the issues you may have to deal with, and increased your confidence and ability in helping women to make choices that are right for them, their families and their babies.

You should now be able to:

1. Understand the definition of informed choice and can explain how it applies to your own role in pregnancy and newborn screening.

2. Explain the different terms used to describe processes to achieve informed choice (eg, shared decision-making, informed decision-making, and supported decision-making).

3. Understand the distinction between informed choice and informed consent.

4. Discuss the legal and ethical implications of informed choice in pregnancy and newborn screening.

5. Acknowledge individual variations in parents’ desire to make choices or seek professional recommendations.

Adapted from Screening Choices: A resource for health professionals offering antenatal and newborn care: http://cpd.screening.nhs.uk/screeningchoices