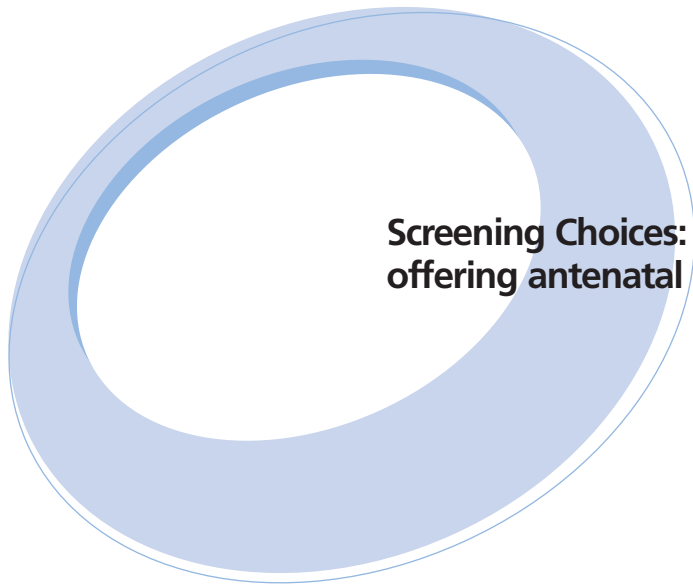


**Screening Choices: A resource for health professionals offering antenatal and newborn care**

**Unit Understanding genetics**

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

This resource has been prepared by

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Cambridge

and

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Cambridge

On behalf of the  
UK National Screening Committee

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## Understanding genetics

# Creating a workbook

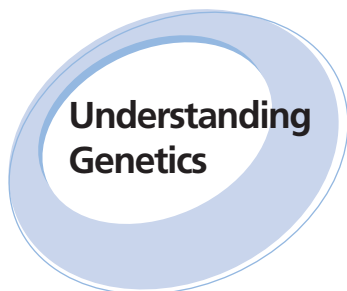


Recording your learning as you work through this unit will enable you to use it as evidence of your professional development. We have developed a workbook for this unit that repeats the activities to help you to structure your work. You can either print out the workbook if you prefer to work on paper,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

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

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

To create your workbook go to Toolbox, click on **Understanding Genetics** then click On-screen workbook or Printed workbook, depending on how you intend to use it.



## Introduction

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

This unit explores some basic aspects of genetics and its implications in antenatal and newborn screening. It aims to help you to develop the knowledge and skills to explain some basic genetic concepts to parents in the context of introducing them to the possibility of genetic screening, and some awareness of the wider implications of genetic applications, both now and in the future. As genetics develops in many areas of healthcare, much of the knowledge will also support you more widely in your professional practice.

### Learning outcomes

After studying this unit you will be able to:

- Understand the basics of human genetics
- Discuss the role of genetic factors in health and disease
- Explain the mode of inheritance in single-gene (Mendelian) disorders (*dominant, recessive and sex-linked recessive*) and provide examples of specific conditions
- 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
- Understand how genetic services and antenatal diagnosis services relate to each other
- Discuss the developments in genetics for future healthcare.

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

## Activity 1



Use your workbook to do this activity

### Self-assessment

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

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

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

#### The self-assessment

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

1. I understand the basics of human genetics.
2. I can discuss the role of genetic factors in health and disease.
3. I can explain the mode of inheritance in single-gene (Mendelian) disorders (dominant, recessive and sex-linked recessive) and provide examples of specific conditions.
4. I 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. I understand how genetic services and antenatal diagnostic services relate to each other.
6. I can discuss the developments in genetics for future healthcare.

#### Identifying your skills and knowledge

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

The purpose of this unit is to help you to develop your understanding of basic genetics as it relates to antenatal and newborn screening. However, it also discusses a range of conditions that are not currently part of national screening programmes. This is to help you to develop a broader understanding of human genetics and genetic inheritance. It will also help you to answer some general questions about genetics you may be asked by women and their partners in the course of consultations about antenatal and newborn screening.

## What is 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 antenatal stage (eg, Down's syndrome and sickle cell disorders) and for the newborn baby (eg, phenylketonuria). This unit 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. For the details of these individual conditions in the national screening programmes you can refer to the specific educational materials that support these programmes (these can be accessed in the Toolbox). It also offers sources of further information.

The term 'genome' means all the genetic material carried in the body. The human genome includes the complete set of genes contained in the human body. Apart from identical twins each individual's genome is unique.

### National screening programmes for genetic conditions (2005)

#### Antenatal screening programmes

- Down's syndrome
- Haemoglobin disorders (sickle cell disorders, thalassaemia disorders): to be available across the UK by the end of 2005

#### 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

#### Pilot newborn screening programme

- Medium chain acyl-CoA dehydrogenase deficiency (MCADD): pilot programmes in some areas

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 unit. 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.

Details on using genetic services and short clinically-oriented articles for health professionals are available on the National Electronic Library for Health (NeLH) website <http://libraries.nelh.nhs.uk/genepool>

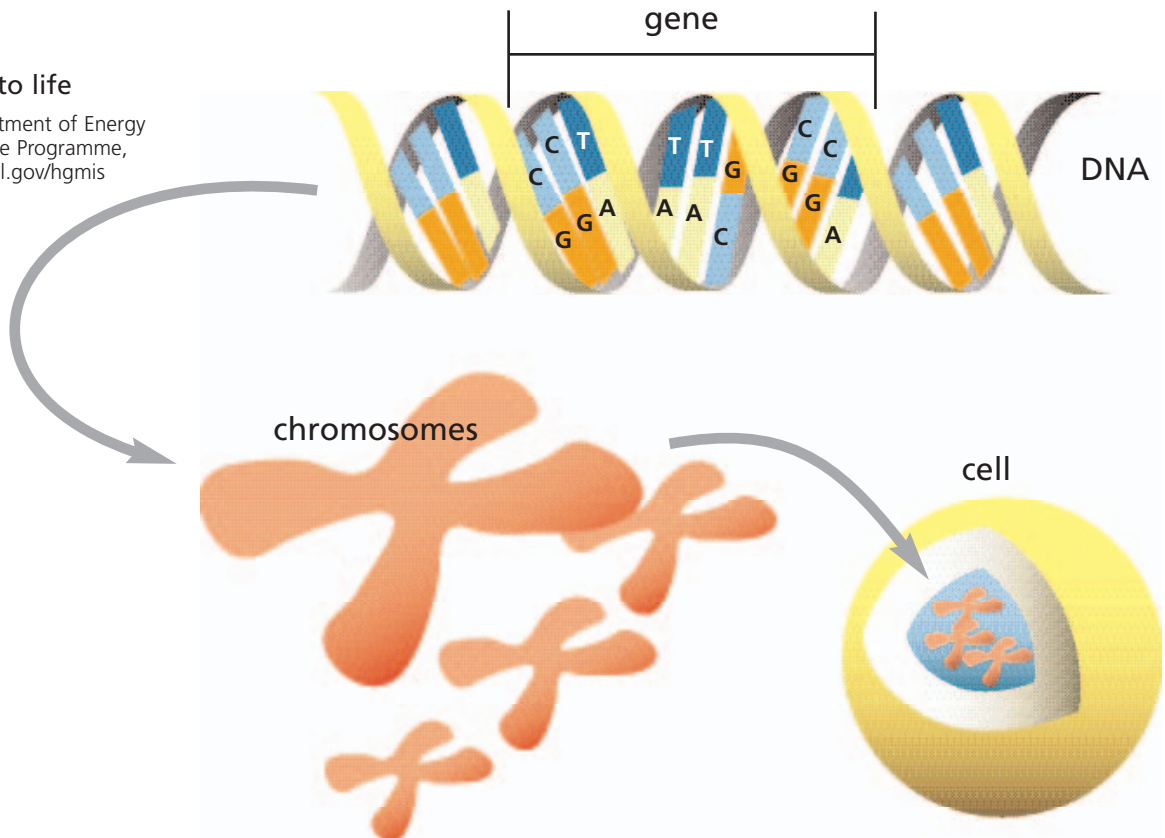
## Genes, chromosomes and DNA

As you work through this unit you will 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, that 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).

Figure 1.  
From DNA to life

From US Department of Energy  
Human Genome Programme,  
<http://www.ornl.gov/hgmis>



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 medical 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 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 (page 7) 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

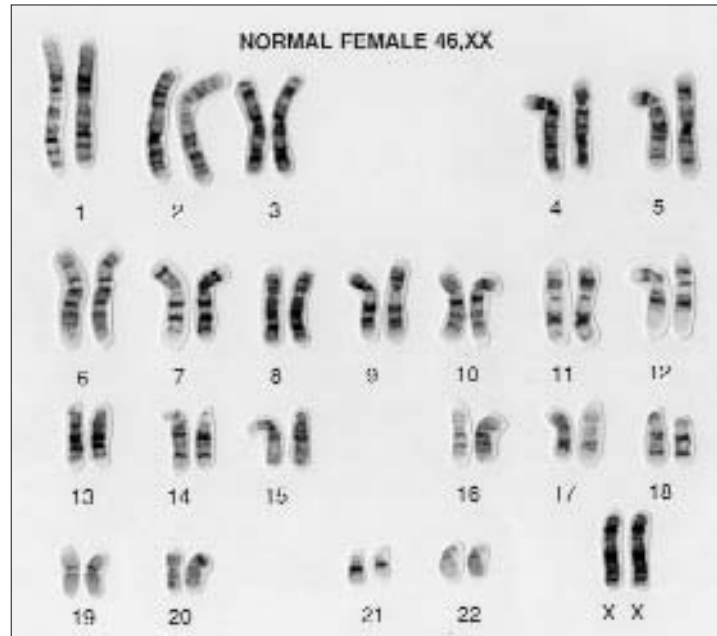
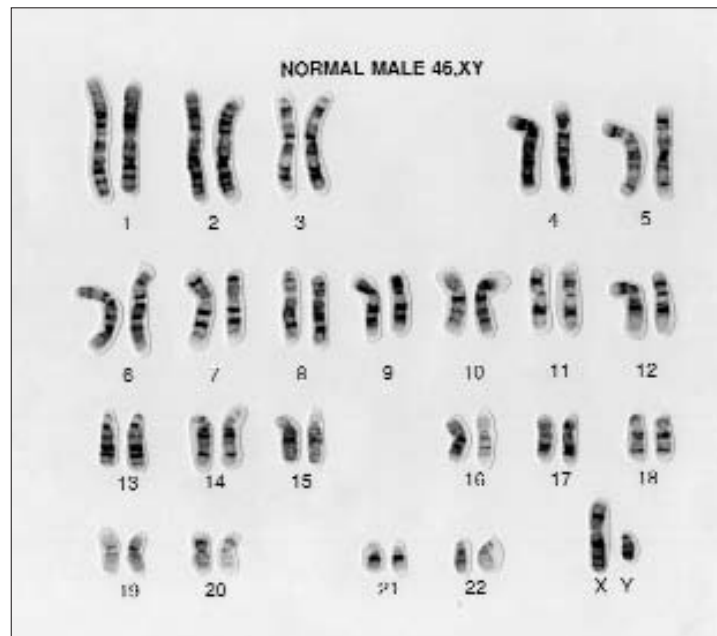


Figure 3.  
A male karyotype  
with the usual 23  
pairs of  
chromosomes



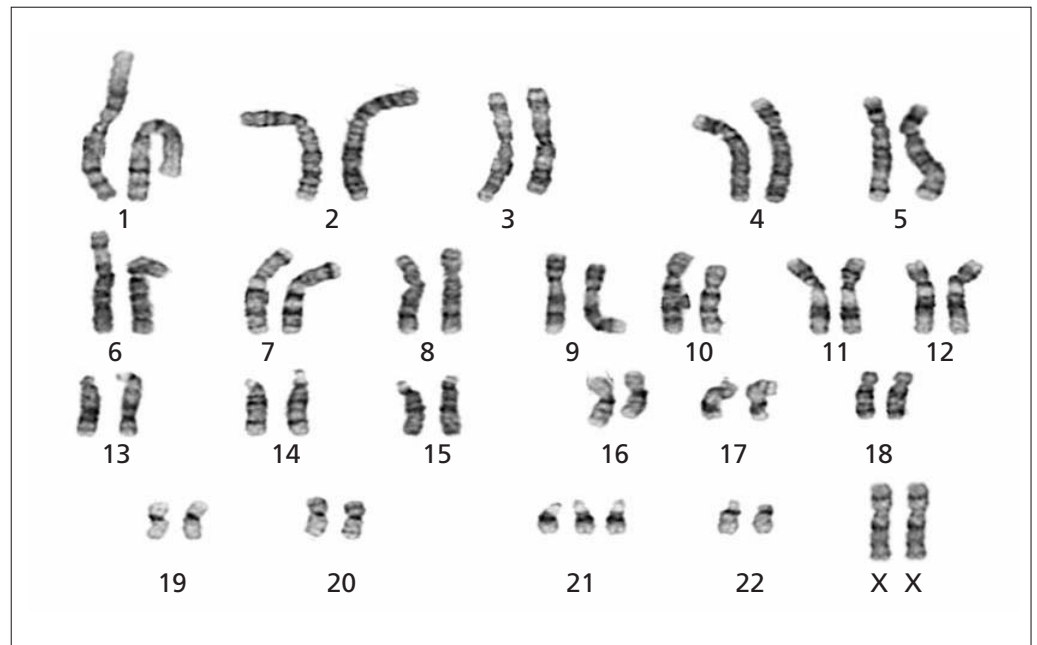
The chromosomes in each matched pair within the karyotypes are similar in shape, size and patterning. Individual chromosomes are similar in all humans, even if some genes on them give the cells of the body very different instructions from one human to another. We inherit one chromosome in every pair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

Figure 4.  
Karyotype of a  
female with  
Down's syndrome



There is more on Down's syndrome in the unit *Screening in antenatal and newborn care* and on the Down's Syndrome Screening Programme website:  
<http://www.nelh.nhs.uk/screening/dssp/home.html>

### 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).

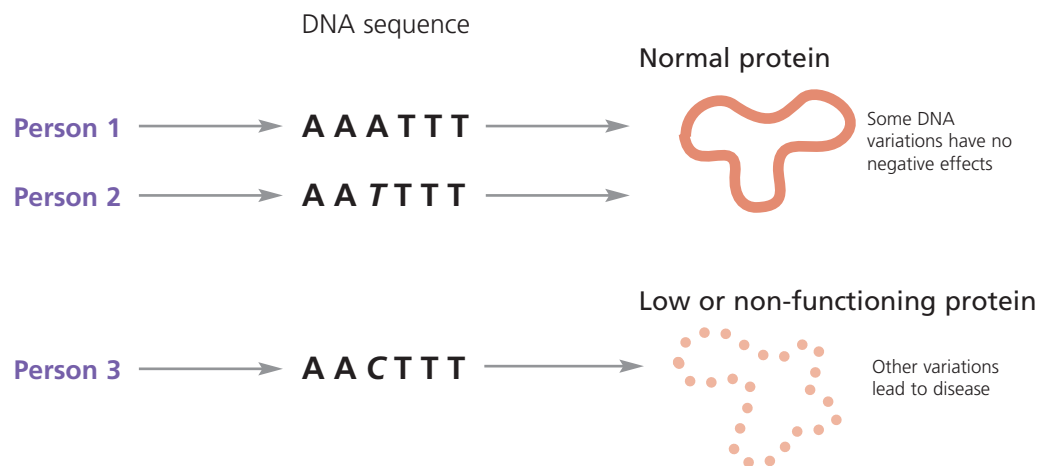
Here is an example of just one of the genes contained on chromosome 11:

**betaglobin gene:** This gene has a role in producing the protein haemoglobin, which transports oxygen in the blood. The haemoglobin disorders (sickle cell disorder and thalassaemia) are associated with alterations in this gene.

*There is more information on sickle cell and thalassaemia on the Sickle Cell Society website (<http://www.sicklecellsociety.org>) and the UK Thalassaemia Society website (<http://www.ukts.org>).*

Figure 5.  
**The effects of altered genetic codes**

Adapted from US Department of Energy Human Genome Program, <http://www.ornl.gov/hgmis>



In Figure 5, the genetic code for the instructions that make proteins is made up of a four-letter alphabet – A, G, C and T (see box below) – where each letter represents one of the four chemical units (bases) which form part of the DNA molecule. A single letter

(base) out of place, a 'simple spelling mistake', in the genetic code can result in a malfunctioning (dysfunctional) protein. This may then result in a condition such as sickle cell disorder, phenylketonuria (PKU) or medium chain acyl-CoA dehydrogenase deficiency (MCADD). The spelling mistake in the genetic code is known as a mutation or alteration.

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.

### 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.

# 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.

### Risk factors

For most diseases or conditions, epidemiological research has identified a range of risk factors that put people at greater or lesser risk. For most diseases, this list of factors includes such things as lifestyle (eg, diet) or the external environment (eg, exposure to radiation) and a family history of the condition.

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

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 diseases

### 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 risk of genetic disease to themselves or their family. The patterns of inheritance vary according to whether autosomes (non-sex chromosomes) 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 8). 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 Down's syndrome on the Down's syndrome screening programme website: <http://www.nelh.nhs.uk/screening/dssp/home.htm>

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**.

Table 1 lists the most common conditions found in newborn babies that are caused by an increased or decreased number of chromosomes. While some cause severe problems and usually lead to death in infancy, in others, life expectancy can be normal.

Table 1.

Syndrome	Abnormality	Birth prevalence per 10,000 births	Lifespan (years)
Down's	Trisomy 21	15	40+
Edwards'	Trisomy 18	3	<1
Patau's	Trisomy 13	2	<1
Turner	Monosomy X	2 (female births)*	Normal
Klinefelter	XXY	10 (male births)*	Normal
XXX	XXX	10 (female births)*	Normal
XYY	XYY	10 (male births)*	Normal

\* Whilst we know the birth prevalence from newborn surveys, the vast majority of these individuals will never be diagnosed if they are not prenatally detected when karyotyping is done to detect Down's syndrome.

You can find out more about Down's syndrome on the Down's Syndrome Association website (<http://www.downs-syndrome.org.uk>), Edwards' and Patau's syndromes on the Support Organisation for Trisomy 13/18 website (<http://www.soft.org.uk>), XXX and XXY syndromes on the Contact A Family website (<http://www.cafamily.org.uk>) and Klinefelter syndrome on the Klinefelter Syndrome Support Group website (<http://klinefeltersyndrome.org>).

Activity 2 asks you to consider Down's syndrome, as this is the most common chromosomal anomaly, and is the only one for which there is a national screening programme.

## Activity 2

### Chromosome anomaly

Core

1. Read the details on Down's syndrome given below.
2. Rewrite the details in your own words or discuss the information with a colleague or friend.



Use your workbook to do this activity

#### Down's syndrome

- Down's syndrome usually occurs sporadically.
- Most people with Down's syndrome have an extra, whole chromosome 21 in all cells.
- The extra chromosome 21 is acquired during the formation of the sex cells (eggs/ova or sperm), most commonly in the egg.
- Standard trisomy 21 is not inherited so it is not necessary to test parental chromosomes. There is an increased maternal age-related risk for Down's syndrome.
- Down's syndrome causes learning disabilities and can cause health problems, but some people are more severely affected than others.

Mother's age at time of baby's birth	Risk of baby being affected with Down's syndrome
15	1 in 1,500
30	1 in 800
45 and over	1 in 50 and greater

(Figures from the Down's syndrome screening programme website: <http://www.nelh.nhs.uk/screening/dssp/home.htm>)

Advanced

Use the information above and the Down's Syndrome Screening Programme website (see link above) to think through how you would describe the underlying genetic basis for Down's syndrome to a woman considering screening who asks more about it. Can you find any resources that would help?

## Feedback

Core

The information presented about Down's syndrome gave you the basic facts about the condition. Generating your own description from the facts or discussing them with a colleague should have supported your ability to communicate and use the information. This will help you to explain basic information about Down's syndrome to women and their partners and families.

Advanced

It is very likely that some of the women and families you work with will want more information. This may be related to a range of subjects such as the genetics of Down's syndrome, the screening and/or diagnostic tests they are being offered, risk and risk factors or how the syndrome can affect people. Understanding all these aspects of Down's syndrome and the screening programme will help you to ensure these people are able to make informed choices about their screening options.

A range of sources of information in the Toolbox could be helpful, either in enabling you to explain genetics or the other issues more clearly to parents, or to enable them to find out more for themselves if they prefer. They may also find support organisations helpful as they make their decision.

Some women and their partners and families may not feel that they need additional information about Down's syndrome. This may be because they already know about it, or are clear about whether or not they want to accept the offer of screening – or they may simply not want any more information. While it is their decision whether or not to accept more information, it is important to discuss this to ensure they are making an informed choice.

---

### **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 risk 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).

### **Using your understanding of the genetic basis of disease**

Your developing knowledge and understanding of the genetic basis of disease will help you to enable women to make informed choices.

*There is more on parents' perspectives on antenatal and newborn screening in the unit **The parent perspective on screening**.*

Activity 3 aims to help you develop your understanding of genetic conditions you encounter in your clinical practice, and your ability to discuss these with women and their partners. However, it is vital to understand the limits of your knowledge and your role, and to refer on to more specialist professionals where this is appropriate.

## Activity 3

### Genetic disorders

Core

Write down (or discuss with a colleague) three disorders that have a genetic basis that you have encountered in your antenatal and/or newborn screening practice, and describe briefly the underlying genetic anomaly.

Advanced

You might be asked by women or their families about particular genetic conditions, or be working with a woman with a particular condition and need to know what the underlying alteration is. You need to be able to research these conditions and explain the underlying genetic basis in basic terms to the woman. Choose one condition from each of the three groups below. Use the websites listed and/or your own research to complete the table in your workbook, which displays:

- The type of disease/disorder
- The factor(s) associated with these genetic diseases (eg, alteration in a gene)
- The main effects of the disease and source(s) of your information (ie, websites, books, article details)
- Sources of further information and support for parents.

#### 1. Alteration in a gene – single-gene disorders (eg, cystic fibrosis, sickle cell disorder, beta-thalassaemia)

##### General

National Electronic Library for Health Clinical Genetics Specialist Library:  
<http://libraries.nelh.nhs.uk/genepool>

##### Cystic fibrosis

BUPA Health Information:

[http://hcd2.bupa.co.uk/fact\\_sheets/Mosby\\_factsheets/Cystic\\_fibrosis.html](http://hcd2.bupa.co.uk/fact_sheets/Mosby_factsheets/Cystic_fibrosis.html)

Cystic Fibrosis Trust: <http://www.cftrust.org.uk>

Newborn Screening Programme Centre:

<http://www.newbornscreening-bloodspot.org.uk>

##### Sickle cell disorder and thalassaemia

Newborn Screening Programme Centre:

<http://www.newbornscreening-bloodspot.org.uk>

Schoolscience:

<http://www.schoolscience.co.uk/content/5/biology/mrc/7/page1.html>

Sickle Cell Society: <http://www.sicklecellsociety.org>

Sickle Cell and Thalassaemia Screening Programme:

<http://www.kcl-phs.org.uk/haemscreening/publications.htm>

UK Thalassaemia Society: <http://www.ukts.org>

#### 2. An alteration in the number of chromosomes – chromosome anomalies (eg, Down's syndrome, Turner syndrome or Klinefelter syndrome)

##### General

National Electronic Library for Health Clinical Genetics Specialist Library:  
<http://libraries.nelh.nhs.uk/genepool>

##### Down's syndrome

Down's Syndrome Association: <http://www.downs-syndrome.org.uk>

Down Syndrome Health Issues: <http://www.ds-health.com/trisomy.htm>



Use your workbook to do this activity

3 continued

**3** *continued*

Down's Syndrome Screening Programme:

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

National Institute of Child Health and Human Development:

<http://www.nichd.nih.gov/publications/pubs/downsyndrome/down.htm#TheChromosomal>

**Klinefelter syndrome**

Klinefelter Syndrome Association UK: <http://www.ksa-uk.co.uk>

Klinefelter Syndrome Support Group: <http://www.klinefeltersyndrome.org>

**Turner syndrome**

Turner Syndrome Support Society: <http://www.tss.org.uk>

**3. The presence of several gene variants (that increase disease risk) and their interactions with environmental factors – multifactorial disorders**

(eg, spina bifida, diabetes mellitus, coronary heart disease)

**General**

Wellcome Trust Polygenic and Multifactorial Diseases:

<http://www.wellcome.ac.uk/en/genome/genesandbody/hg06b010.html>

**Coronary heart disease**

British Heart Foundation: <http://www.bhf.org.uk>

**Diabetes**

British Diabetic Association: <http://www.diabetes.org.uk>

**Spina bifida**

Association for Spina Bifida and Hydrocephalus:

<http://www.asbah.org/Spina%20Bifida/Support.html>

**Feedback****Core**

You may find that some women and families decline any explanation of genetic anomalies. This should be respected and documented. However, some will want at least a simple explanation, and it is important to be able to give this. While you may need to refer to information resources for detailed explanations, being able to give a simple explanation is likely to increase their confidence in your practice. Generating your own description should help you to retain the information about conditions you encounter.

**Advanced**

If screening or diagnostic tests reveal a risk or the presence of a genetic condition in their fetus or baby, many parents will want more detailed explanations of their underlying genetic basis. You need to be able to give them information in a way that is meaningful to them and, where necessary, refer them to sources of further information. You need to know how to access more detailed information and this activity will have helped you not only to find information on three conditions you encounter, but how to access information on conditions more generally. Presenting the information in table form as you have in this activity will serve as a useful aide memoire when you discuss the conditions with women and families.

For more information about genetic testing services go to:

National Electronic Library for Health Clinical Genetics Specialist Library:

<http://libraries.nelh.nhs.uk/genepool>

Guy's and St Thomas' NHS Foundation Trust Genetics Centre:

<http://www.guysandstthomas.nhs.uk/page2040.htm>

For more information about genetic testing go to:

Lawrence Berkeley National Library, What is Genetic Testing?:

<http://www.lbl.gov/Education/ELSI/Frames/genetic-testing-f.html> (an American website)

## Inheritance patterns

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

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 risk 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. Activity 4, which is at core level only, aims to help you to develop your understanding of genetics and inheritance patterns.

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.

### Activity 4

#### Genetics and inheritance patterns

Read through *The Genetics Primer* found on the Genetics, Disease and Dentistry website. This is a good introduction for all health professionals – it gives clear, concise information on genetic inheritance (launched in 2004 by the National Coalition for Health Professionals in Genetics NCHPEG).

[http://www.nchpeg.org/dental/genetic\\_primer/tableofcontent.html](http://www.nchpeg.org/dental/genetic_primer/tableofcontent.html)

You can link to and explore whole website from <http://www.nchpeg.org>

### Feedback

*The Genetics Primer* is a simple, clear introduction to genetics, and is useful for all health professionals. If the women and families you work with ask for sources of information on genetics, you may find it helpful to refer them to *The Genetics Primer* and/or some of the websites listed in the Toolbox.

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

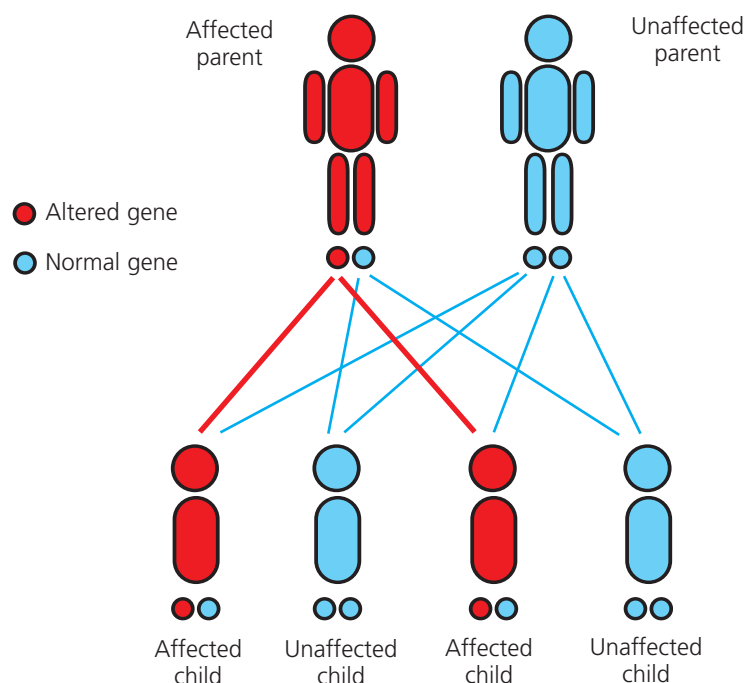
Huntington's disease is an autosomal dominant disorder associated with the presence of an autosomal dominant allele. For the disease to develop in an individual he or she needs only one copy of the altered allele. Because the condition is associated with non-sex chromosomes, males and females are equally 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 (alteration) occurred when the egg or sperm was being made.

Some single-gene disorders are congenital, apparent at birth or soon after (such as achondroplasia). Some, however, like Huntington's disease begin in later life and an individual carrying the alteration may have no symptoms until he or she reaches middle age.

Figure 6.  
Autosomal dominant inheritance (Mendelian pattern of inheritance)



Some other examples of autosomal dominant conditions include:

- Marfan syndrome
- Neurofibromatosis
- Familial adenomatous polyposis (a form of colon cancer).

### In-depth research

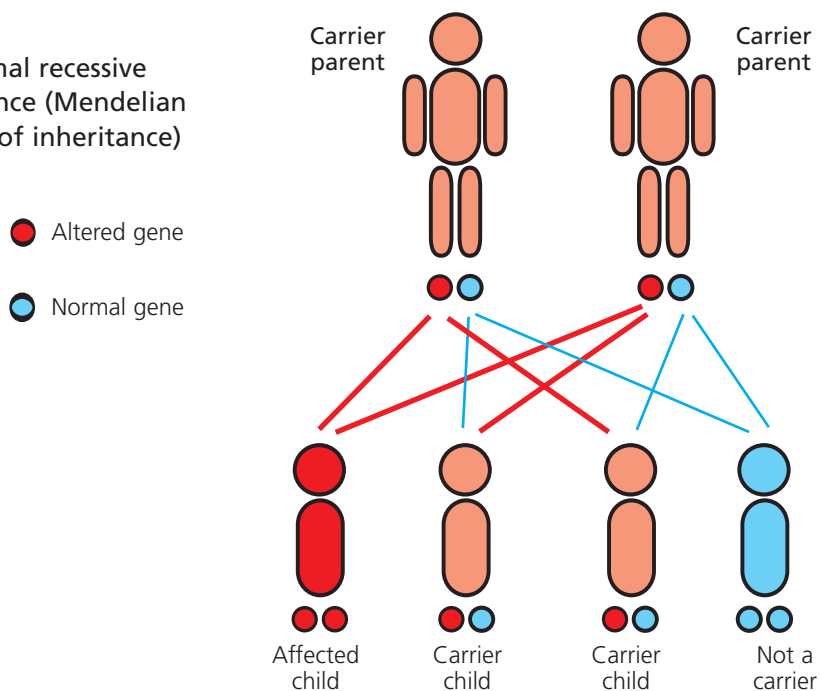
Use the university of Washington's GeneTests site (<http://www.geneclinics.org>) to search by disease for comprehensive information. The site has a useful illustrated glossary.

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.

### 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.

Figure 7. Autosomal recessive inheritance (Mendelian pattern of inheritance)



### 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.

For further information on CF, see the Cystic Fibrosis Trust website (<http://www.cftrust.org.uk>). For further information on screening for CF, see the UK Newborn Screening Programme Centre website (<http://www.newbornscreening-bloodspot.org.uk>) or the materials produced by the programme – *Newborn Blood Spot Screening in the UK: Health Professional handbook and Newborn Blood Spot Screening in the UK: Training resources*.

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.

**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 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>

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.

If you want to find out more about autosomal recessive inheritance and disease:

- Refer to the Public Health Genetics Unit diseases database:  
[http://www.phgu.org.uk/info\\_database/diseases/index.html](http://www.phgu.org.uk/info_database/diseases/index.html)
- Complete an exercise on the genetics of cystic fibrosis using the *BMJ* student website: [http://www.studentbmj.com/back\\_issues/1103/education/405.html](http://www.studentbmj.com/back_issues/1103/education/405.html)

### 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 (Figure 8, page 20).

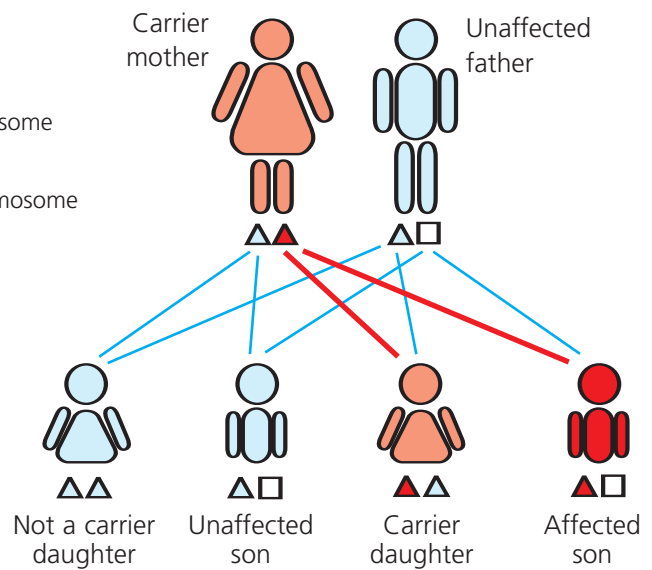
**Duchenne muscular dystrophy** is a condition in which there is progressive muscular weakness because muscle cells break down due to a defect in dystrophin a single important protein in muscle fibres. It affects only boys (with extremely rare exceptions), who first show difficulty in walking between 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.muscular-dystrophy.org>

### Why is there no male-to-male transmission of sex-linked genetic disorders?

Because the altered gene is located on the X chromosome and, by definition, male (XY) children have received the Y chromosome and not the X chromosome from their father (XY).

Figure 8.  
Sex-linked inheritance where the mother is a carrier and the father is unaffected



Other examples of sex-linked conditions are:

- Haemophilia
- Hunter syndrome
- Colour blindness
- Fragile X syndrome.

### Sex-linked inheritance

Further details on sex-linked inheritance can be found at:  
<http://www.cafamily.org.uk/inherita.html>

Activity 5 aims to help you to develop your understanding of inheritance patterns.

## Activity 5

### Understanding inheritance patterns

Core

Write down in your own words a brief outline of single-gene (Mendelian) inheritance, including what you see as the main elements.

Advanced

A mother with whom you are discussing screening for cystic fibrosis does not understand how her baby could have the genetic condition since there is no history of the disease in either her or her partner's family.

How would you explain autosomal recessive inheritance to her so that she can understand that her baby can still be at risk?



Use your workbook to do this activity

## Feedback

Core

Your outline of single-gene inheritance should have contained the following elements.

The disease or condition is:

- Caused by an alteration in a single gene
- Inherited in distinct patterns that can be observed in families.

The patterns of inheritance can be:

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

This depends on whether:

- The mutation needs to be present on one or both copies of the gene for the disease or condition to occur
- The autosomes or the sex chromosomes are affected.

Advanced

Cystic fibrosis is an autosomal recessive condition, so people will usually be unaware that they carry the altered gene that can result in their children having condition. This is because the other gene of the pair is working sufficiently to mean we do not get the disease or condition. So in explaining why her baby might be at risk of the condition you could tell the mother that we all carry recessive altered genes that we are not aware of, and if a baby inherits two copies of the altered gene that is not working he or she will get the disease. For this to happen both parents must carry the altered gene and the child must have inherited both copies of it. If both parents are unaffected carriers, they may pass on either the altered gene or the unaltered one to the baby. The chance for CF is 1:2,500 births, one in 25 of the general population is a carrier. Where both parents are carriers the risk of having an affected baby is 1:4 in each pregnancy.

You may use Figure 7, which shows autosomal recessive inheritance patterns, to help the woman to understand this.

## Genetics, health services and disease prevention

Clinical and laboratory genetic services play an increasing role today in diagnosing genetic disease, or in identifying people who have alterations or anomalies in their genetic make-up that mean that they or their children are at risk of genetic disorders. At present, health services are almost entirely restricted to considering patients at risk of single-gene or chromosomal disorders. Working with the genetics of multifactorial disease such as spina bifida, diabetes, coronary heart disease or rheumatoid arthritis is still confined to research.

Specialist genetic services are provided from regional centres and include the work of clinical geneticists, genetic counsellors, and laboratory services. Specialists are highly trained and experienced in the diagnosis of genetic disorders, risk assessment and supporting people in decision-making – a process known as non-directive genetic counselling. The service is offered to individuals, parents and family members where there is concern about genetic disease either for themselves or their children.

### Genetic services and testing

*For more information of genetic services go to:*

National Electronic Library for Health Clinical Genetics Specialist Library:  
<http://libraries.nelh.nhs.uk/genepool>

Guy's and St Thomas' NHS Foundation Trust Genetics Centre:  
<http://www.guysandstthomas.nhs.uk/page2040.htm>

*For a summary of genetic testing go to:*

Australian Centre for Genetics Education Factsheets:  
<http://www.genetics.com.au/factsheet/19.htm>

Lawrence Berkeley National Library, What is Genetic Testing?:  
<http://www.lbl.gov/Education/ELSI/Frames/genetic-testing-f.html>

In screening programmes, we try to identify individuals at higher risk of a disease or condition so they can be offered options to reduce risk of the condition or its severity. The principles of this are discussed further in the unit *Screening in antenatal and newborn care*. In the antenatal and newborn screening programmes this involves some tests for genetic conditions, although the tests themselves may not involve examination of the genetic material itself (DNA or chromosomes). Identification in the screening programmes also takes place in several stages. Initial screening tests pick out individuals who are at higher risk of a condition, who are then offered further tests that can provide a diagnosis.

Just because a condition is genetic does not necessarily mean it has been inherited. For example, in most cases of Down's syndrome the extra chromosome 21 is a new event; the anomaly has not been inherited from the parent and is unlikely to happen again. However, for other diseases, the discovery of a genetic anomaly in a parent, newborn baby or fetus can have implications for other family members or future children. The details of this will depend on the way in which that disease or condition is inherited.

We can test for disease or genetic anomalies that can lead to increased risk of disease in newborn babies, in adults (in the case of screening programmes this can be one or both parents), and in the fetus during pregnancy. The implications of each of these for disease

prevention are different, and are described in more detail in the unit *Screening in antenatal and newborn care*.

The newborn blood spot test (the heel prick which tests for PKU, CHT, sickle cell disorders and cystic fibrosis) 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 antenatally. If this test is positive the father can also be

offered a test. If both parents are carriers there is a risk 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 antenatal 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.

### Case example

This example illustrates the importance of having some basic understanding about a sex-linked recessive condition. It also shows that prevention can be about being prepared for a baby who is affected with haemophilia.

Elizabeth, aged 22, was 14 weeks into her first pregnancy. She was adopted as a child, but two years ago she traced her brother Clinton, who was also adopted. They had kept in occasional contact, although they were not close. Two days previously, Elizabeth telephoned Clinton to wish him happy birthday and tell him she was pregnant. Clinton asked her if she had ever been tested for haemophilia, as he had the condition. This was the first time Elizabeth had even heard of the condition, and she visited her GP in some distress, asking whether she too might have haemophilia and whether her baby might have it.

Elizabeth's GP explained what the condition was and how it was inherited, and told her she could be tested to see if she was a carrier. If she was found to be a carrier, she could opt to have a diagnostic test to check whether her baby was affected. At first Elizabeth said she would not want to know whether or not her baby was affected, because she would not consider having a termination. However, the GP explained that if the baby was affected, diagnosis would enable the maternity service to make special arrangements for the delivery to minimise the risks to herself and her baby. Elizabeth changed her mind and decided to have the tests.

## 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 (eg, 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:

- Many disabled people themselves believe that having a screening programme to look for serious disease in the antenatal period devalues those with the conditions, the pleasure that they can get out of life and the contribution they can make to society.
- There are feelings of stigmatisation in having a disease or condition that 'runs in the family' and sometimes parents who are carriers of conditions, or those who are affected themselves, have a sense of guilt at having passed it on to their children.
- There are issues of confidentiality relating to whether or not other family members might need to be informed if they or their children are also at risk of the disease, because of what we know about how it is inherited.
- Sometimes testing parents for carrier status can reveal the fact that the woman's partner could not be the father of the child. This situation has to be handled very carefully by those with specialist understanding and skills.

Activity 6, which is at core level only, asks you to consider some of the implications of genetic testing and genetic conditions, looking in particular at cystic fibrosis.

## Activity 6

### Cystic fibrosis – implications for other family members

Read the background information below, from the NHS Direct online health encyclopaedia and then consider and/or discuss the questions with a colleague.

**Cystic fibrosis** is an autosomal recessive condition in which an altered version of a protein called cystic fibrosis transmembrane conductance regulator (CFTR) is produced. This is responsible for transporting salts and water across the cell membranes. This means that in certain parts of the body, the secretions lack water, becoming thick and sticky. The lungs, pancreas, intestines and other organs tend to get clogged up with thick, sticky mucus. Symptoms include poor weight gain, chest infections, coughs, abnormal stools and salty sweat.

1. Parents of a child with CF have a 25% chance (one in four) of recurrence for each child conceived. How do you think this knowledge may affect their decision to have another child?
2. Imagine you are working with parents who already have one child who is affected and are sure they could not cope with another, but would like to have a second child. What sort of options might be available to them, and how could you help?



Use your workbook to do this activity

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## Feedback

This activity should have helped you to consider some of the dilemmas that parents face when there are genetic conditions in the family.

1. In answering this question you might have included how the scientific knowledge combined with the parents' social, moral and religious beliefs shapes the decision that they may make. You could explore some of the options presented in Abramsky and Chapple (2003).
2. You could tell the parents in general terms that there are options available to help that would include being able to tell if the baby is affected early in pregnancy, so that they can consider whether or not to proceed with the pregnancy. One further option which some couples find preferable, is to use in vitro fertilisation to create an early embryo and test this at the stage of a few cells, only then implanting a non-affected embryo in the uterus. This is known as pre-implantation genetic diagnosis. It is a very specialist area and the couple would need to be referred for further specialist advice. Another option with recessive conditions is to consider artificial insemination by donor if the father is a carrier or affected.

The parents will need to consider how they and their first child might feel about this and whether or how they might discuss it with him or her.

*You can find out more about CF on the Cystic Fibrosis Trust website (<http://www.cftrust.org.uk>) and the National Electronic Library for Health Clinical Genetics Specialist Library: <http://libraries.nelh.nhs.uk/genepool>*

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## 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.

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.

### Further information

School Science Updates, Cystic Fibrosis: The quest for a cure:  
<http://www.schoolscience.co.uk/content/5/biology/mrc/3/page5.html>

**Gene therapy:**  
Human Genome Project Information, Gene Therapy:  
[http://www.ornl.gov/sci/techresources/Human\\_Genome/medicine/genetherapy.shtml#whatis](http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetherapy.shtml#whatis)

**Stem cell research:**  
Wellcome Institute: <http://www.wellcome.ac.uk>  
Stem Cell Institute: <http://www.stemcells.cam.ac.uk>

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.

## Conclusion

Working through this unit 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 antenatal and newborn screening for genetic conditions. Where situations are outside your knowledge it is vital to refer parents on to specialist genetics services.

Activity 7 is another self-assessment that should help you to check whether you have met your learning outcomes and identify any areas where you may need to increase your skills or knowledge. Other units in this programme, materials from the other antenatal and newborn screening programmes or specialist sources of genetics information may help you with this.

### Activity 7

#### Self-assessment

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

1. I understand the basics of human genetics.
2. I can discuss the role of genetic factors in health and disease.
3. I can explain the mode of inheritance in single-gene (Mendelian) disorders (dominant, recessive and sex-linked recessive) and provide examples of specific conditions.
4. I 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. I understand how genetic services and antenatal diagnostic services relate to each other.
6. I can discuss the developments in genetics for future healthcare.

#### What now?

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

## Focus activity



Use your workbook  
to do this activity

## Explaining genetics

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

Catherine has a newborn baby, her first child, and declined an offer of antenatal screening. She has used the internet to find out about the Newborn Screening Programme and sickle cell disorder, and asks: 'If my partner and I have no family history of sickle cell disorder, why do you want to screen our baby for this condition?'

How would you explain to Catherine why her baby is being offered screening for sickle cell disorder as part of the Newborn Bloodspot Screening Programme?

### Think about:

- The inheritance pattern of sickle cell disorder
- The potential benefits of screening in newborn care
- The ethical, legal and social implications for Catherine and her family

## Reference

Abramsky, L., Chapple, J. (2003) *Prenatal Diagnosis: The human side* (2nd edn). Tewkesbury: Nelson Thornes.

## Further reading

Firth, H.V., Hurst, J.A., Hall, J.G. (advisory editor) (2004) *Oxford Desk Reference of Clinical Genetics*. Oxford: Oxford University Press.

Harper, P.S. (2004) *Practical Genetic Counselling* (6th edn). London: Arnold.

Skirton, H., Patch, C. (2002) *Genetics for Healthcare Professionals: A lifestage approach*. Oxford: BIOS Scientific Publishers.

## Websites

Association for Spina Bifida and Hydrocephalus:  
<http://www.asbah.org/Spina%20Bifida/Support.html>

Australian Centre for Genetics Education: <http://www.genetics.com.au>

BUPA Health Information:  
[http://hcd2.bupa.co.uk/fact\\_sheets/Mosby\\_factsheets/Cystic\\_fibrosis.html](http://hcd2.bupa.co.uk/fact_sheets/Mosby_factsheets/Cystic_fibrosis.html)

Children Living with Inherited Metabolic Diseases: <http://www.climb.org.uk>

Contact A Family (for information on XXX and XXY syndromes and other genetic disorders): <http://www.cafamily.org.uk>

Cystic Fibrosis Trust: <http://www.cftrust.org.uk>

Down's Syndrome Association: <http://www.downs-syndrome.org.uk>

Down's Syndrome Health Issues: <http://www.ds-health.com/trisomy.htm>

Down's Syndrome Screening Programme:  
<http://www.nelh.nhs.uk/screening/dssp/medical.html>

Genetics Primer: [http://www.nchpeg.org/dental/genetic\\_primer/tableofcontent.html](http://www.nchpeg.org/dental/genetic_primer/tableofcontent.html)

Guy's and St Thomas' NHS Foundation Trust Genetics Centre:  
<http://www.guysandstthomas.nhs.uk/page2040.htm>

Human Genome Project Information:  
[http://www.ornl.gov/sci/techresources/Human\\_Genome/home.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml)

Klinefelter Syndrome Association UK: <http://www.ksa-uk.co.uk>

Klinefelter Syndrome Support Group: <http://klinefeltersyndrome.org>

Lawrence Berkeley National Library, What is Genetic Testing?:  
<http://www.lbl.gov/Education/ELSI/Frames/genetic-testing-f.html>

Muscular Dystrophy Campaign: <http://www.muscular-dystrophy.org>

National Coalition for Health Professionals in Genetics: <http://www.nchpeg.org>

National Electronic Library for Health Clinical Genetics Specialist Library:  
<http://libraries.nelh.nhs.uk/genepool>

National Human Genome Research Institute: <http://www.genome.gov>

National Institute of Child Health and Human Development:  
<http://www.nichd.nih.gov/publications/pubs/downsyndrome/down.htm#TheChromosomal>

Public Health Genetics Unit: <http://www.phgu.org.uk>

Sanger Institute: <http://www.sanger.ac.uk>

Schoolscience: <http://www.schoolscience.co.uk>

Sickle Cell Society: <http://www.sicklecellsociety.org>

Sickle Cell and Thalassaemia Screening Programme:  
<http://www.kcl-phs.org.uk/haemscreening/publications.htm>

Stem Cell Institute: <http://www.stemcells.cam.ac.uk>

Student BMJ Genetics of Cystic Fibrosis Quiz:  
[http://www.studentbmj.com/back\\_issues/1103/education/405.html](http://www.studentbmj.com/back_issues/1103/education/405.html)

Support Organisation for Trisomy 13/18 (Edwards' and Patau's syndromes):  
<http://www.soft.org.uk>

Turner Syndrome Support Society: <http://www.tss.org.uk>

UK Newborn Screening Programme Centre: <http://www.newbornscreening-bloodspot.org.uk>

UK Thalassaemia Society: <http://www.ukts.org>

University of Washington, GeneTests: <http://www.geneclinics.org>

Wellcome Trust: <http://www.wellcome.ac.uk>

Wellcome Trust Polygenic and Multifactorial Diseases:  
<http://www.wellcome.ac.uk/en/genome/genesandbody/hg06b010.html>

Unit: Understanding genetics

# Certificate of completion

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

I am able to:

- Understand the basics of human genetics
- Discuss the role of genetic factors in health and disease
- Explain mode of inheritance in single-gene (Mendelian) disorders (dominant, recessive and sex-linked recessive)
- 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
- Understand how genetic services and antenatal diagnostic services relate to each other
- Discuss the developments in genetics for future healthcare.

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

Signature

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Name

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Job title

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Date

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\* Delete as appropriate