

FOUR GENETIC CONDITIONS

TEACHERS PLEASE NOTE

This activity is designed to support science teachers who have already covered some aspects of inheritance with their Key Stage 4 students. It is not intended as an introduction to genetic conditions.

It is important to be aware that some students in the class may themselves have a genetic condition, or be a carrier, or have a relative who is affected. Sensitivity is required to avoid putting such students under stress.

OBJECTIVE

To increase awareness and understanding of four genetic conditions (Haemophilia, Cystic Fibrosis, Huntington's Disease and Down's Syndrome) and of different patterns of inheritance

SUGGESTED AGE RANGE

14—16 year olds who have completed the genetics component within National Curriculum Science or its equivalent.

CURRICULUM LINKS

Science

National Curriculum Science (Double) at KS4, Life Processes and Living Things, Section 4: Variation, inheritance and evolution.

'Pupils should be taught: f. that some diseases can be inherited.'

Scottish Certificate of Education, Biology — Standard Grade, Topic 6: Inheritance. Learning outcomes, General Level — 'Describe one example of a human condition caused by a chromosome mutation e.g. Down's Syndrome.'

MATERIALS NEEDED

Photocopy the statements about the four conditions on pages 133-140 and cut them up (if you photocopy each set onto different colour paper or card, they can be more easily sorted after the lesson for use again with other groups). Shuffle each complete set of statements well before placing in an envelope. You will need one complete set of cut-up statements per group of four students.

Photocopy Activity Sheets A and B (one of each per group of four students).

Four Genetic/Chromosomal Condition Cards are available to support this Activity. They are: Genetic Condition Card: Haemophilia
Genetic Condition Card: Huntington's Disease
Genetic Condition Card: Cystic Fibrosis
Chromosomal Condition Card: Down's Syndrome.

USING THE ACTIVITY WITH STUDENTS

Ask students to work in groups of three or four. Read through Activity Sheet A as a whole class and make sure that everyone understands the task. Give each group a copy of Activity Sheet B and a complete set of cut-up statements. If you wish, you can also give each group a copy of the four Genetic/Chromosomal Condition Cards listed above to help them with the task of sorting out the statements. Set a time limit for the activity.

ACTIVITY SUPPORT FOUR GENETIC CONDITIONS :

ACTIVITY SHEETS FOR STUDENTS

ACTIVITY SHEET A

Throughout our adult life, millions of our cells divide each second and every minute we produce thousands of miles of newly copied DNA.

Sometimes mistakes happen during the copying process. These mistakes can be limited to one letter of the genetic code or sometimes to whole chromosomes.

A change in the structure of DNA is known as a genetic mutation. We all have some mutations in our genes. Mutations can happen spontaneously when the sperm or egg are forming. They can also be inherited.

Not all mutations show an effect. However some can seriously affect a person's health and quality of life.

There are more than 4500 conditions and disorders which are caused by faulty genes or changes to whole chromosomes. You may already have learned about some of these conditions. This Activity focuses on four:

- Haemophilia
- Huntington's Disease
- Cystic Fibrosis
- Down's Syndrome

WHAT YOU DO

Take Activity Sheet B. You will see that it is divided into four squares.

Share out the statements you have been given equally amongst the students in your group. Each statement tells you something about one of the genetic conditions listed on Activity Sheet B. Some statements give you information. Others give you an idea of how some people with the condition think and feel. Your task is to decide which statement goes with which condition.

Take it in turns to read a statement and try to place it on the correct square on Activity Sheet B. If there are any statements that you cannot place on Activity Sheet B, put them on one side.

When everyone has placed as many statements as they can on Activity Sheet B, discuss any which are left over before trying to place them on the Activity Sheet too. Then compare each condition; are there positive and negative aspects to all of them?

ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

ACTIVITY SHEET B



HAEMOPHILIA

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This is an inherited and lifelong blood condition where an essential blood clotting factor is either partly or completely missing. This causes a person with this condition to bleed for longer than normal. Cuts and grazes are no great problem as a little pressure and a plaster are usually enough to stop the bleeding.

The main problem is internal bleeding into joints, muscles and soft tissues.

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A person who is mildly affected might not find out that they have this condition until they have an operation or a tooth out at the dentist, or injure themselves badly and go on bleeding for longer than most people would. But severely affected children are often diagnosed within the first year of life. One early symptom is that babies with the condition bruise very easily when they start to crawl. Unfortunately, doctors sometimes wrongly think that the bruises are the result of physical abuse by the parents.

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It is important to remember that this condition is inherited and not infectious (so you cannot 'catch' it). It is known as an X-linked (or sex-linked) recessive condition because the faulty gene which leads to the condition is situated on the X chromosome (one of the sex chromosomes).

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The faulty gene for this condition is carried on the X chromosome. Females have two X chromosomes (XX), males an X and a Y chromosome (XY).

All males inherit one X chromosome from their mother. If this X chromosome contains the faulty gene, the boy will have the condition, but if he inherits the other X chromosome, he will not. Females who inherit the X chromosome containing the faulty gene do not have the condition. They are carriers and can pass the gene onto any children they have, but the gene on their other X chromosome works normally (although sometimes a woman who is a carrier can be mildly affected).

When a father has this condition, he has a faulty X chromosome and will pass this to a daughter who will become a carrier. This will always happen because, unlike a woman, he has only one X chromosome so there is no possibility of passing on a working copy of the gene.

In the unusual circumstances of a woman carrier and a man who has this condition having a child together, it is possible for a daughter to be affected. This is because the daughter may inherit two faulty X chromosomes, one from each parent. This is extremely rare and in nearly all cases it is males who have the condition.

ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

HAEMOPHILIA continued

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People can be mildly, moderately or severely affected. The degree of severity does not vary within individuals, so if someone is mildly affected, this will always be so. For a mildly affected person, problems usually only occur after a severe injury or an operation

If moderately affected, problems are usually related to some injury such as a knock or deep cut. However, severely affected people often have regular internal bleeding into joints, muscles and tissues. No injury is necessary. Children tend to bleed more than adults

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People with this condition can be in pain when a bleed occurs, especially if it is not treated promptly by an injection of the missing clotting factor. Over time, regular and untreated bleeding causes severe joint damage which can eventually lead to mobility problems. A person with this condition should never take aspirin as this thins the blood and can make bleeding worse.

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HAEMOPHILIA

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Treatment for people who are severely affected is usually by replacement of the missing clotting factor. This is done through injections on a regular basis to help prevent bleeding, or injecting at the time a bleed occurs. Regular preventative treatment (i.e. two or three

times a week) helps the blood to clot. It also minimises the likelihood of bleeding and of pain and long-term joint damage too. There is no permanent way of replacing or increasing the clotting factor level.

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The clotting factor is derived from donated blood. Until 1986, people with this condition were affected by contamination of blood products leading to widespread infection with HIV and Hepatitis C.

Approximately 1,200 of them have been infected with HIV. Tragically, over half of these people have died as a result of developing AIDS. These bereavements have severely affected families due to the loss of sons and fathers.

Over 3,000 people with this condition have been infected with Hepatitis C which affects the functioning of the liver. The long-term future of infected individuals is uncertain as the disease often takes some years to develop.

Since 1986, when heat treatment of blood products started, no new infections have occurred. A 'recombinant' clotting factor — where the missing factor is synthetically produced — is now being introduced to help avoid the risk of contamination through viruses found in human blood.

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This is a rare condition affecting about 10,000 males in the UK. Females are so rarely affected that no statistics have been kept.

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ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

HAEMOPHILIA

'When I was about 11 months old, I cut my gum and it didn't stop bleeding for three days so I was taken to hospital for some tests and they discovered that I had the condition. No-one else in my family is affected.

I have an internal bleed about once every two weeks.

I can usually tell when it's happening and I give myself an injection to stop the bleeding. I'm 17 now and I've been doing my own injections since I was about 10. I have a freezer bag which I carry my stuff in. I take it to college and sometimes to friends' or I just come home if I have a bleed. After a bleed I may need to use a wheelchair to take the weight off my feet and rest my joints, but I can still get in to college because they have wheelchair access. I suppose the worst bits are if I have to come away when I'm at friends' because of a bleed, and the times when I'm off my feet for a bit and I'd rather be out enjoying myself. I don't see myself as disabled. People who I spend a bit of time with know I have the condition, and my college lecturers do too. But I've lived with it all my life and I'm not treated any differently because of it.'

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HUNTINGTON'S DISEASE

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People at risk of getting this inherited condition can develop it at any age, but it is most common for them to start to show signs of the disease later in life (i.e. between the ages of 30 and 50). Although people use the words 'illness' and 'disease' to describe this condition, it is important to remember that the condition is inherited and not infectious (so you cannot 'catch' it). The only way you can develop the disease is if you have inherited a faulty copy of the gene from one of your parents. Unfortunately you will develop the disease even if you inherit one working copy of the gene as well as the faulty gene. This is because the faulty gene is dominant.

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If the condition does not show itself until a person is middle-aged, other family members may not be aware that they are at risk. Imagine that a parent or grandparent develops the disease in their sixties. Suddenly, their children are faced with the fact that there is a 50% (1 in 2) chance that they have inherited the faulty gene and will develop the disease. And not only them, but possibly their children too. This can be very difficult for families.

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About 1 in every 10,000 babies born in the UK will have inherited the faulty gene which leads to this condition.

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This condition is caused by a years and gets progressively emotionally (they may the affected person changes walking, eating and talking mutation in a gene which affects the way the brain functions. The illness usually begins with mild symptoms spread over a number of worse until the person dies, perhaps of pneumonia or heart failure. Because the disease destroys brain cells, become depressed, moody or aggressive), physically (their movements become jerky and uncontrolled and become difficult) and in the end, they cannot think or reason clearly.

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ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

There is no known cure for this condition at the moment. But sympathetic care is important and medicines can sometimes help to reduce the jerky movements.

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You may have heard the word 'chorea' (pronounced ker -ree -er) used to describe this condition. Chorea is the Greek word for dancing and describes the jerky movements made by affected people. But the word 'disease' is used more often than 'chorea' these days because the jerky movements are not the only symptoms.

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HUNTINGTON'S DISEASE

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In families where there is a person with this condition, it is possible for other family members who think they might have inherited the faulty gene to have a test. After counselling, a blood sample is taken and the DNA in it is analysed. Waiting for the result can be very stressful. A negative result means that you do not have a copy of the faulty gene. This means that neither you nor your children will develop the disease.

A positive result means that you have inherited the faulty gene and will develop the disease, but the test can't tell you when that will happen. The test helps some families, particularly when the results are negative or where people feel that not knowing is worse than knowing. But lots of people do not have a test because they do not want to know if they are going to get a disease for which there is no cure.

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'I am 29 years old. I first found out about the condition when I was '13. My father suffered from it and died at the age of 44. My grandmother and my uncle had it too. My father became ill with obvious symptoms when he was about 38 years old. I was 14 at the time. I didn't realise what it was until we were told when I was 15. It was awful — we knew nothing. All I knew was that my father was odd ; falling all over the place, very stubborn and could be violent. I , or I should say we , knew nothing about the illness. Mum left Dad but I couldn't , I loved him no matter what. When he died I experienced nightmares that you just could not imagine. My life carried on. I had a little boy who is now seven years old. Then earlier this year I decided to have the test. I cannot tell you how wonderful the lady was who counselled and supported me right through this time. At first I didn't know what to do. To have the test? Not to have the test?

I was so confused. It took a lot of courage and a lot of thinking — very deep thinking.

It finally came , the morning my test result was due and I felt so much anguish I didn't know what I was doing. I didn't want my boyfriend there. I just wanted to be on my own. I received the result and it was what I expected - positive. So I have the faulty gene and at some time in the future I will develop the disease. All I can say is that I feel positive about my future and my little boy's. I want to tell everyone to be strong and to remember that people die of worse things. I will be strong, I have to be and I want to be. I have a wonderful partner who is understanding, loving and supportive. He is there for me and, fingers crossed, he always will be. I have a wonderful son who is intelligent and loving and I have lots of good understanding friends around me.'

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ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

'Being a carer can be a very putting my own needs difficult and demanding job, even when you are caring for someone you love. Looking after my wife, who has this condition, often means second and it can seem selfish to even think about myself. But everyone who is caring, no matter what age they are, needs help sometimes. Many young carers keep silent about their work but talking to others about how you feel can help you to realise you are not alone. It isn't easy, and a lot can depend on finding the right person to talk with.'

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CYSTIC FIBROSIS

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This is the UK's most common life-threatening inherited disease. Every week another five babies are born with the condition.

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This condition affects mainly the lungs and the pancreas (the pancreas produces vital enzymes needed for digesting food). Thick, sticky mucus builds up in these organs causing infections and damage in the lungs and making digesting food difficult. The male reproductive system can also be affected.

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If you know a person with this condition, you may notice that they cough a lot (this cough is not infectious — you cannot 'catch' it). They may also be small for their age even though they have to eat lots of high-calorie foods.

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To stay well, people with this condition need to do physiotherapy and breathing exercises to clear the mucus from their lungs. This usually takes up a lot of time at least once or twice a day (or more particularly if they are unwell). They also take enzymes with each meal in powder or capsule form to help them digest their food. Many will take prescribed vitamin supplements and oral antibiotics too, so they may be taking about 20 pills with each meal. A good diet and regular exercise can also help them to stay well. But most people with the condition also need to make regular hospital visits to check how they are or for further treatment.

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This condition is most often found in people of European origin. In the UK it is estimated that 1 person in 25 is a carrier (that's about 2 million people or 4% of the population).

Genes come in pairs, one from each parent. A person who inherits one faulty copy of a recessive gene and one working copy is known as a carrier. Most carriers do not know that they have the faulty gene because the working copy functions normally. So they are not ill or affected in any way by the condition, but they can pass the faulty gene on to any children they may have.

ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

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It is important to remember that this condition is inherited and not infectious (so you cannot 'catch' it). It is known as a recessive condition because a person has to inherit two copies of the faulty gene - one from each parent — to be affected.

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Each (and every) time two carrier parents of this condition have a child, there is:

- A 25% (1 in 4) chance that they will have a child with this condition
- A 50% (1 in 2) chance that they will have a child who is an unaffected carrier like themselves
- A 25% (1 in 4) chance that they will have a child who does not have the condition and is not a carrier.

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CYSTIC FIBROSIS

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There is no cure for this condition. But the gene has been found and researchers are beginning to understand more about how it works. So new genetic techniques can be used to make better drugs to treat the condition, and as treatments improve people with this condition are living longer into adult life. Gene therapy is another exciting development. Doctors are looking at ways of getting working copies of the gene into the lungs of people with the condition and of making sure that the genes go on working when they are there. Gene therapy is not yet a reality but holds hopes for the future

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A simple mouthwash sample or blood test can be used to find out if a person is a carrier of this condition. This test accurately detects up to 80% (but not all) of the mutations in the gene. There are companies who offer this test privately — you send off a sample and a fee and get the results through the post. Some people say that postal testing is a bad thing because it does not include pre- and post-test counselling to help interpret the results. Others think that carrier testing should be offered free on the NHS to everyone who is thinking of having children.

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'It was a shock when I was turned down for health insurance. I was 20, self-employed and didn't really see my condition as a problem. But life went on, I started my business, got married and bought a home, although getting a mortgage was difficult because they saw me as a bad risk.

Things have improved on the treatment front ; this year I've had three courses of intravenous antibiotics at home, which is much better than having to go into hospital every time I get a chest infection which won't clear up. But I wish I'd known before I got married that men with this condition are usually infertile. It's just lucky that I've got an understanding wife because not being able to have children is hard.

I know that my health has got worse over the last five years because I get more infections than I used to. So life's not a bed of roses. But it's not that bad either because I have a wife, a house and a job. In lots of ways I'm not really different from anyone else. I do think about dying ; some days more than others, but I don't let it dominate me. I just don't make plans too far into the future!'

ACTIVITY SUPPORT: FOUR GENETIC CONDITIONS

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DOWN'S SYNDROME

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A person with this condition is born with 47 chromosomes in each body cell instead of the usual 46.

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On average, three babies are born with this condition each day in the UK.

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If a woman with this condition became pregnant, there would be a high chance that the baby would inherit the condition (particularly if the baby's father had the condition too).

But most women who have a baby with this condition do not have the condition themselves. At some point when the egg or sperm is made, or perhaps at fertilisation, an unusual cell division takes place. This results in an extra copy of chromosome number 21 in all the baby's cells. There is no known reason why this occurs. It happens by chance and can happen to anyone. A few people are born with the condition because they have inherited a chromosome abnormality, called a translocation, from one of their parents.

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This condition is not an illness or a disease so it is not appropriate to talk about a cure. Many people with the condition enjoy a healthy life and can live to between 50 and 70 years old.

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Some of the physical features which you might see in a person with this condition can include eyes which appear to slant slightly upwards and outwards, a round head and rather flat face, and perhaps rather short fingers and small stature. But this does not mean that everyone with the condition will look the same. They will have their own, and some of their family's, physical characteristics too. People with this condition will also have learning difficulties. But these vary, and you cannot judge a person's ability from their appearance.

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Physiotherapy, speech therapy and extra teaching at home and in school can help children with this condition to reach their full potential

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It is fairly common for people with this condition to be born with a heart defect and sight problems.

Difficulties with hearing can also occur and will probably vary throughout the person's life.

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DOWN'S SYNDROME

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It is possible for any woman, whether or not she has the condition, to have a test when she is pregnant which will tell her if her unborn baby has the condition. One common test is called amniocentesis. It is usually carried out in the fourth month of pregnancy and involves taking a sample of the fluid which surrounds the baby in the womb. It is almost 100% accurate in diagnosing the condition but the test also causes about one in every hundred women to lose the baby through miscarriage. It is important for all women who decide to be tested for this and other conditions during pregnancy to talk with a trained health professional/counsellor before and after the test. This can help people to understand the test and to think about how they will feel, and what will they do, when they get results.

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In the general population, older women, especially those over the about age 35, have an increased chance of having a baby with this condition (the age of the father appears to be less important).

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‘When I was young, my parents taught me to do most of the things which other children do – like feed and dress myself, read, talk, tie my shoelaces and so on – but it took me longer to learn than many children. When I was in my teens and still at school, I went half a day a week for about two terms to the local college for further education. I did computing and word processing and a bit of shop work and basic administration. When I left school I went straight to a residential college for the disabled. I started there on the 10th January 1989 – I can remember the date exactly! After I left college at Easter 1992 I got a work experience job through the MENCAP Pathway scheme as a large head office. I did the internal and external mail in the post room and some work in the advertising department. I really enjoyed my two and a half years there.

By the time I had left home and was living in a residential place to be more independent. It was okay at first but then things started to slide and I got into my own world of late nights and TV and secret eating. I started to feel unwell. I was 21 and the youngest there and it just wasn’t the right place for me. I lost my job because I couldn’t get up in the mornings. Eventually I came back home to live until I find a more suitable group home.

I’ve been back at home for about two years My interest now is looking at the community and at what people do in it. I write letters asking people to send information to me so I get a lot of mail and have a lot of reading to do. I sometimes deliver local free newspapers. I go by train to my gran’s every Tuesday and do housework for her. We cook lunch together and some weeks she asks me to do her shopping. And I work once a week in a small office doing photocopying mostly. My ambition is to go to America and Australia, as I am really interested in accents and how other people live.

I don’t see myself as different – I know I have the condition and I just get on with my life as a normal person doing what anyone does really.

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