

Genetic Testing - Discussion Continuum

Introduction:

As DNA techniques and our knowledge of the human genome develop it is becoming increasingly easy to test DNA for indicators of genetic disorders.

This resource, suitable for KS4 and post 16 audiences, gives students the opportunity to investigate genetic disorders and asks them to consider a number of statements and situations concerning the social and ethical issues surrounding genetic testing.

This is a very flexible resource that could be used as part of wider work on the topic or as a lesson in its self. This resource has been regularly used with groups of 12 students; they discuss the issues raised by each statement and agree where each card should go between 'agree' and 'disagree'. Larger groups could use the resource to have a free discussion of the topic or you could use formats that require the students to work more formally or in smaller groups. Please see our Formats for Discussion information sheet for more suggestions.

Contents:

The resource consists of:

- An 'agree' and a 'disagree' card in case you would like to use the format described above and set up a 'value continuum'. This is where the students have to decide, as a group, and taking into account everyone's opinion on the matter, where they should place the card on a continuum between agree and disagree.
- 12 Discussion Cards, which include a statement on some aspect of genetic testing and if appropriate some information on the genetic condition they refer to.
- 6 Information Sheets, containing more detailed information on the genetic conditions referred to by the discussion cards

Page	Content
1	Introduction and contents
2	Agree/disagree cards
3 – 6	Discussion cards
7 – 12	Information sheets

A red button with rounded corners and a black border. The word "DISAGREE" is written in white, uppercase, sans-serif font in the center.

DISAGREE

A green button with rounded corners and a black border. The word "AGREE" is written in white, uppercase, sans-serif font in the center.

AGREE

Genetic testing and counselling should be free on the NHS, even if it costs the taxpayer more money.

People who carry the genes for terrible diseases like Huntington's should not be allowed to have children. They should adopt instead.

Huntington's Disease (dominant) affects the central nervous system, it causes increasing coordination and memory problems, mood changes and eventually death. Symptoms usually start around 30 – 50 yrs old but this is highly variable.

Mortgage and loan companies should have access to your genetic records - they don't want to lend money to someone who might get sick or die.

Genetic testing kits for common diseases like cystic fibrosis should be available over the counter at chemists.

Cystic Fibrosis (recessive) causes thick salty mucus to build up in the pancreas, lungs and bowel. Life expectancy depends on the severity of symptoms, but the average is 25-30yrs. There are a variety of treatments available for the symptoms but no cure.

I know that I am a carrier of Duchenne muscular dystrophy. I should be able to use IVF techniques to make sure that my next baby is a girl.

Duchenne muscular dystrophy (recessive – but is X linked so males always display symptoms) causes muscular weakness. Symptoms start between ages 1 and 3, sufferers are normally wheelchair bound by 12 and life expectancy is around 20yrs.

All newborn boy babies should be tested for Duchenne muscular dystrophy. That way parents would be forewarned before they have any more children.

Duchenne muscular dystrophy (recessive – but is X linked so males always display symptoms) causes muscular weakness. Symptoms start between ages 1 and 3, sufferers are normally wheelchair bound by 12 and life expectancy is around 20yrs.

I have FHC and there is a chance that my children will have too. I should be able to get them tested so that I can change their diets if necessary.

Familial hypercholesterolemia (dominant) results in abnormally high blood cholesterol levels and hence an increased risk of heart disease. The mean age of onset of heart disease is 40 for male sufferers; 50-55 for female. This can be treated with drugs and a special diet.

I am 11 and my Grandad has Huntington's disease. My dad doesn't want me to be tested but I do. I should be able to make the choice for myself.

Huntington's Disease (dominant) affects the central nervous system; it causes increasing coordination and memory problems, mood changes and eventually death. Symptoms usually start around 30 – 50 yrs old but this is highly variable.

Pre-implantation and prenatal genetic testing should be banned. You should just accept and love children as they are.

My wife and I have achondroplasia. We should be able to use IVF techniques to select a child who has achondroplasia too.

Achondroplasia (dominant). Achondroplasia is also known as short limbed dwarfism. Individuals with heterozygous achondroplasia have normal IQ and lifespan and an average adult height of four foot. Homozygous individuals however will be stillborn.

My first child has Sanfilippo syndrome. When I get pregnant again I should be able to have prenatal testing for the disease.

Sanfilippo Syndrome (recessive) results in the build up of waste in cells, causing cell damage. Progressive mental deterioration occurs between 5 and 10yrs, children become hyperactive and disruptive, movement and speech are then lost and death occurs in the mid teens.

All airline pilots should be tested for FHC. This will improve passenger safety.

Familial hypercholesterol aemia (dominant) results in abnormally high blood cholesterol levels and hence an increased risk of heart disease. The mean age of onset of heart disease is 40 for male sufferers; 50-55 for female. This can be treated with drugs and a special diet.

Information file

Cystic fibrosis

Population frequency

1 in 2500 in Northern Europe, 1 in 25 people carry the CF gene.

Pattern of inheritance

Recessive, caused by a single-gene mutation on chromosome seven. An affected parent has a 25% chance of having an affected child, but only if both parents are carriers. Carriers show no symptoms.

Phenotype

The gene involved codes for a protein called CFTR (Cystic Fibrosis Transmembrane Conductance Regulator). This is bound to membranes in the lungs and gut and transports salt and water into and out of cells. In CF the protein does not function properly and thick salty mucus builds up in the lungs, pancreas and bowel.

Symptoms

Because the mutation can occur at different points in the gene, severity of symptoms is very variable. Problems include lung damage, infections, blockage of the bowels and difficulties digesting food. Life expectancy depends on the severity of symptoms but the average is 25-30 years.

Treatment

Pancreatic enzymes can be taken to help improve digestion. Mucus in the lungs can be shifted using daily physiotherapy, and drugs can help reduce inflammation and destroy bacteria. Lung transplants are possible, and trials of gene therapy have shown some success.

Possibility of genetic testing

Some health authorities use genetic testing to screen all newborn babies for CF. Mouth swab tests are available for people who have an affected relative. It is also possible to carry out pre-natal testing.

Information file

Familial hypercholesterolaemia (FHC)

Population frequency

1 in 500

Pattern of inheritance

Dominant, caused by various mutations on chromosome 19. An affected parent has a 50% chance of having an affected child.

Phenotype

The gene for FHC codes for specific receptors that remove excess cholesterol from the blood. This means that sufferers have an abnormally high blood cholesterol level.

Symptoms

The high blood cholesterol level leads to an increased risk of coronary heart disease. If left untreated, the mean age of onset of heart disease is 40 for male sufferers and 50-55 for females.

Treatment

Cholesterol levels can be controlled with a combination of drugs and a special diet. This means that once diagnosed, people with FHC can reduce their risk of heart disease to near normal levels.

Possibility of genetic testing

Diagnosis can be made either by measuring blood cholesterol levels or genetic testing.

Information file

Achondroplasia

Population frequency

1 in 26 000.

Pattern of inheritance

Dominant, caused by a single-gene mutation. Over 80% of children born with achondroplasia have no history of the disorder in the family - there has been a spontaneous new mutation. An affected parent has a 50% chance of having an affected child.

Phenotype

The gene involved codes for a protein called fibroblast growth factor (FGFR3). This helps to control bone growth. Affected individuals have abnormal bone growth, especially in the limbs. Another name for achondroplasia is short-limb dwarfism.

Symptoms

Individuals with heterozygous achondroplasia have normal IQ and lifespan. The average adult height is about four foot. Small bones can lead to some complications including middle ear infections, backache, respiratory problems and hydrocephalus (fluid collecting round the brain). Babies with achondroplasia tend to take longer to reach developmental milestones such as sitting and walking.

Treatment

It is possible to increase limb length using surgery, but this is uncommon. Careful monitoring of children can reduce the risk of spinal damage or hydrocephalus.

Possibility of genetic testing

Achondroplasia can be detected in the womb using ultrasound. If both parents are affected by achondroplasia there is a 25% chance of their child being homozygous for the achondroplasia gene. If this is the case the child will be stillborn or will die soon after birth. There is a DNA test for this condition.

Information file

Huntingdon disease

Population frequency

1 in 18 000

Pattern of inheritance

Dominant, caused by a single gene on chromosome 4. An affected parent has a 50% chance of having an affected child.

Phenotype

The gene codes for a protein called Huntingtin. At present the action of this protein is not fully understood. In some way the faulty gene results in progressive damage to nerve cells in the brain.

Symptoms

Symptoms usually develop when people are between 30-50 years old, but this is very variable. Early signs include involuntary movements, concentration and memory problems and mood changes. These get more severe later in the illness. Death occurs on average about 17 years from the onset of symptoms, but this is again highly variable.

Treatment

At present there is no cure for Huntingdon disease, but medication, speech therapy and special diets can help treat the symptoms.

Possibility of genetic testing

Genetic tests are available - they will tell you if you have the gene, but will not give an indication of the age at which you will develop symptoms. Prenatal testing is also possible.

Information file

Duchenne muscular dystrophy (DMD)

Population frequency

1 in every 3500 male births

Pattern of inheritance

Recessive, caused by a gene on the X chromosome. X-linkage means that females can be carriers, but males with the faulty gene always display symptoms.

Phenotype

The gene codes for a muscle protein called dystrophin. Without a working copy of the gene, boys suffer progressive muscular weakness. They also have an increase in the enzyme creatine kinase in the blood.

Symptoms

Symptoms usually develop when boys are between 1 and 3 years old. They may have difficulty walking and jumping. Sometimes they may have learning difficulties. By the age of 12, most boys with the disease have to use a wheelchair. The disease also affects heart and diaphragm muscles and average life expectancy of a boy with DMD is about 20 years.

Treatment

At present there is no cure for DMD, but medication can help to alleviate the symptoms.

Possibility of genetic testing

Boys can be tested for DMD in two ways. The first is analysis of the levels of creatine kinase in the blood. The second is a genetic test. It is also possible to carry out a prenatal test for the sex of subsequent children.

Information file

Sanfilippo syndrome

Population frequency

1 in 85 000

Pattern of inheritance

Recessive, caused by a single-gene. An affected parent has a 25% chance of having an affected child, but only if both parents are carriers. Carriers show no symptoms.

Phenotype

The gene involved codes for an enzyme called heparan sulphatase, which breaks down waste mucopolysaccharides in cells. Without a functioning enzyme, cells become progressively damaged as the mucopolysaccharides build up.

Symptoms

Babies start with no or few symptoms, but between the ages of five and ten progressive mental deterioration occurs and children become hyperactive and disruptive. Eventually movement and speech are lost and death occurs in the mid teens.

Treatment

At present there is no cure for Sanfilippo syndrome.

Possibility of genetic testing

Sanfilippo syndrome is so rare that parents tend to find that they are carriers only when they have a child who is affected. However, for subsequent children, they can opt to have chorionic villus sampling during pregnancy to see if the foetus is affected.