B1

Science, Genetics

B1 Genes are found in our chromosomes and parents pass these on to offspring in their sex cells. Different versions of the same gene are called alleles, and these can determine features such as eye colour, and the inheritance of disorders such as cystic fibrosis. DNA You will remember from your Key Stage 3 studies that the nucleus controls the activities of a cell. The instructions for how an organism develops are found in the nuclei of its cells Chromosomes Chromosomes are structures found in the nucleus of most cells. They consist of long strands of a substance called deoxyribonucleic acid, or DNA for short. A section of DNA that has the genetic code for making a particular protein is called a gene. The proteins can either be: * structural proteins such as the ones found in muscles and hair * enzymes, such as proteases and other digestive enzymes Nucleus, chromosome and gene Sex cells and chromosomes Human body cells each contain 23 pairs of chromosomes. Parents pass on their genes to their offspring in their sex cells. * female sex cells are called egg cells, or ova * male sex cells are called sperm Process of fertilisation A pair of chromosomes carry the same genes, in the same place, on the chromosome. However, there are different versions of a gene called alleles. These alleles may be the same on each pair of chromosomes, or different, for example, blue eyes or brown eyes. Sex cells only contain one chromosome from each pair. When an egg cell and sperm cell join together, the fertilised egg cell contains 23 pairs of chromosomes. One chromosome in each pair comes from the mother, the other from the father. Which chromosome we get from each pair is completely random. This means different children in the same family will each get a different combination. This is why children in the same family look

a little like each other and a little like each parent, but are not identical to them. Sex chromosomes A set of chromosomes can be separated from its cell, spread out on a microscope slide and magnified many thousands of times. When stained and photographed, they look like this: Chromosomes from a female Chromosomes from a male The highlighted pair of chromosomes are called the sex chromosomes. The longer sex chromosome is called the X chromosome, the shorter one the Y chromosome. * females are XX * males are XY Determination of gender When sex cells form, the pairs of sex chromosomes (XX and XY) are separated. Remember that females carry XX, males XY. This means: * all normal egg cells produced by a human ovary have an X chromosome * half of the sperm carry an X chromosome, and half a Y So a human baby’s gender is determined by the sperm that fertilises the egg cell. The baby will be a girl if it carries an X chromosome. It will be a boy if the fertilising sperm carries a Y chromosome. Study the animation below to test your understanding of this. Notice that half of the babies should be male, and half female. Individual families often have more, or less, boys than girls, but in a large population there will be roughly equal numbers. This is for the same reason that if you toss a coin many times you will get roughly equal numbers of ‘ heads’ and ‘ tails’. Ideas about science - choosing gender Some societies prefer to have male children. It is now possible to choose the sex of a child using IVF. Some people think parents should be able to choose the sex of their children, especially if they have a child that dies, or have had three or four children of the same sex. Other people think we should not be able to choose, because this could affect the balance of males and females in society, or because

they believe it is against God or nature. Different values - higher only Decisions of this kind are called values. Science can provide information and data, but it cannot answer questions about values. Values often result in different people coming to different decisions. This is why some people think we should be able to choose the sex of our children, while others do not. The Y chromosome - higher The Y chromosome carries a gene called the ‘ sex-determining region Y’, or SRY for short. The SRY gene causes testes to develop in an XY embryo. These produce androgens: male sex hormones. Androgens cause the embryo to become a male. Without them, the embryo develops into a female. Variation Individuals differ in all sorts of ways, even when they are offspring of the same parents. These differences are called variation. Most characteristics, such as height, are determined by several genes working together. They are also influenced by environmental factors. These include: * climate * diet * physical accidents * culture * lifestyle For example, an individual might inherit a tendency to tallness, but a poor diet during childhood will result in poor growth and a shorter individual. Identical twins Identical twins are genetically the same. They are a good example of the interaction between inheritance and the environment. For example, an identical twin who takes regular exercise will have better muscle tone than one who does not. All of the differences that you see between identical twins, for example, in personality, tastes and aptitude, are due to differences in their experiences or environment. Alleles
The chromosomes in a pair carry the same genes in the same places. But there are different versions of the same gene. Different versions of the same gene are called alleles, which is pronounced al-eels. For example, the gene

for eye colour has an allele for blue eye colour and an allele for brown. For any gene, a person may have the same two alleles, or two different ones. Recessive or dominant alleles Alleles may be either recessive or dominant. * A recessive allele only shows if the individual has two copies of it. For example, the allele for blue eyes is recessive. You need two copies of this allele to have blue eyes. * A dominant allele always shows, even if the individual only has one copy of it. For example, the allele for brown eyes is dominant. You only need one copy of it to have brown eyes. Two copies will still give you brown eyes. Individuals A and B have brown eyes - only individual C has blue eyes Only individual C will have blue eyes, because the allele for blue eyes is recessive. Individual A is called a carrier because, even though they have brown eyes, they still carry the allele for blues eyes and can pass this allele on to future generations. Cystic fibrosis Cystic fibrosis (CF) is caused by a recessive allele. In the genetic diagram below, it is written as f. People with CF produce abnormally thick and sticky mucus in their lungs and airways. As a result, they are more likely to get respiratory infections. Daily physiotherapy helps to relieve congestion,
while antibiotics can fight infection. CF also affects the gut and pancreas, so food is not digested efficiently. Inheriting copies of the allele You need to inherit two copies of the faulty allele to be born with CF. If you have just one copy, you are a carrier, but will not experience any symptoms. If two carriers have a child together, there is a one in four chance of it inheriting the disorder. The genetic diagram shows why. Inheritance of cystic fibrosis Huntington’s disorder Huntington’s disorder is caused by a dominant allele, written as H. The symptoms usually develop in middle age, and include

problems with controlling your muscles and forgetfulness. Inheriting copies of the allele You only need to inherit one copy of the faulty allele to have Huntington’s disorder, unlike cystic fibrosis, where you need to inherit both copies. You can inherit Huntington’s disorder if one or both of your parents carry the faulty allele, because it is a dominant allele. You can show inheritance of the disorder using genetic diagrams. In this example, one parent, the mother, carries one copy of the Huntington’s allele. She has the disorder. The father does not carry the Huntington’s allele, so he does not have the disorder. There is a 1: 2, or 50 per cent, chance of them producing a child with the disorder. Note that in an individual family, by chance, all of the children may inherit the disorder, or none at all. Inheriting the Huntington's allele In this example, both parents carry one copy of the Huntington’s allele. Both have the disorder. There is a 3: 4, or 75 per cent, chance of them producing a child with the disorder. Note that in an individual family, by chance, all of the children might inherit the disorder. It is also possible for none of them to inherit the disorder, although this is less likely than in the first example. In this instance, one parent, the mother, carries one copy of the Huntington’s allele. The father carries two copies. Both have the disorder. All of their children will have it, too. Ideas about science - values Scientists are now able to test adults and foetuses for alleles which can cause genetic diseases. However, the scientific information produced raises many issues that science cannot address. For example, should a couple with a one in four risk of having a child with cystic fibrosis take the gamble, or decide not to have any children at all? If a woman becomes pregnant with a child that is going to have cystic fibrosis, should she have

the child, or choose to have an abortion? These are questions about values that science cannot answer. Different people will have different views. Genetic testing can determine whether a person is carrying the alleles that cause genetic disorders. But there are limits to the testing, and the subject raises a number of ethical issues. Genetic tests Genetic testing involves analysing a person’s DNA to see if they carry alleles that cause genetic disorders. It can be done at any stage in a person’s life. There are several types of genetic test, including testing for medical research. Antenatal testing This is used to analyse an individual’s DNA or chromosomes before they are born. At the moment, it cannot detect all inherited disorders. Prenatal testing is offered to couples who may have an increased risk of producing a baby with an inherited disorder. Prenatal testing for Down’s syndrome, which is caused by a faulty chromosome, is offered to all pregnant women. Neonatal testing Neonatal testing involves analysing a sample of blood taken by pricking the baby's heel This is used just after a baby has been born. It is designed to detect genetic disorders that can be treated early. In the UK, all babies are screened for phenylketonuria, congenital hypothyroidism and cystic fibrosis. Babies born to families that are at risk of sickle cell disease are tested for this disorder. Carrier testing This is used to identify people who carry a recessive allele, such as the allele for cystic fibrosis. It is offered to individuals who have a family history of a genetic disorder. Carrier testing is particularly useful if both parents are tested, because if both are carriers there is an increased risk of producing a baby with a genetic disorder. Predictive testing This is used to detect genetic disorders where the symptoms develop later in life, such as Huntington’s

disorder. Predictive testing can be valuable to people who have no symptoms but have a family member with a genetic disorder. The results can help to inform decisions about possible medical care. Limits of genetic testing Genetic tests are not available for every possible inherited disorder. And they are not completely reliable. They may produce false positive, or false negative, results. These can have serious consequences. False positives A false positive occurs when a genetic test has wrongly detected a certain allele or faulty chromosome. The individual or family could believe something is wrong when it is not. This may lead them to decide not to start a family, or to choose an abortion, in order to avoid having a baby with a genetic disorder. False negatives A false negative happens when a genetic test has failed to detect a certain allele or faulty chromosome. The individual or family would be wrongly reassured. This may lead them to decide to start a family or continue with a pregnancy. Gene therapy Gene therapy involves inserting copies of a normal allele into the chromosomes of an individual who carries a faulty allele. It is not always successful, and research is continuing. It is illegal to do this to sex cells, because any changes would be inherited by the individual’s offspring. Instead, gene therapy is used on body cells. It means the individual could pass on their faulty allele to their children, even if they get better themselves. The basic process Gene therapy involves these basic steps: 1. Doing research to find the gene involved in the genetic disorder. 2. Cutting out the normal allele. Special enzymes are used to do this. 3. Making many copies of the allele. 4. Putting copies of the normal allele into the cells of a person who has the genetic disorder. The gene therapy process Problems in the process The main difficulty is usually the

last step. Here are some of the problems: * the alleles may not go into every target cell * the alleles may join with the chromosomes in random places, so they do not work properly * treated cells may be replaced naturally by the patient’s own untreated cells Different methods Different methods are used to get the alleles into the patient’s cells, including: * using fat droplets in nose sprays * using cold viruses that are modified to carry the allele - the viruses go into the cells and infect them * the direct injection of DNA Ideas about science - ethical issues You will need to use your Ideas about science to: * Distinguish questions that can be answered using a scientific approach from those that cannot. For example, science can answer the question, " What are the chances of my child having cystic fibrosis?". However, it cannot answer the question, " Should I have my pregnancy terminated?" * State clearly what the issue in any debate is. For instance, in a debate about treating genetic diseases with gene therapy, some people think that altering our DNA is against nature or God. The ethical issue is whether scientists should be allowed to use gene therapy. * You will also need to summarise the different views that different people might hold. For example, if gene therapy saves lives, it can only be a good thing. Or, we should not use gene therapy because we do not know the long-term outcomes. * Identify arguments that are based on the right decision, ie a decision that produces the best outcome for most of the people involved. So, if a certain type of gene therapy involves a risk of causing harm, but 90 per cent of the people who have the therapy are cured, is it worth taking the risk? * Identify when certain actions are never justified because they are unnatural or wrong. For example, we could wipe out cystic fibrosis in one generation if we made sure

that anyone who was a carrier was not allowed to have children. However, most people would consider this action to be immoral and wrong. Ideas about science - values - higher Access to our genetic profile Some people believe employers should have access to our genetic profile when we apply for a job. In the future, they may be able to use this information to determine how much time we are likely to take off due to illness, and use this information to help decide whether or not to offer us a job. Insurance companies may also want access to our genetic profile. They could use this information to see how long we are expected to live, and maybe either refuse to give us insurance or only provide it at an increased cost. Both of these are examples of values that science cannot address. Genetic testing is an example of what is technically feasible, ie what can be done. But the decisions we take on how to use this information are an example of values, and cannot be ‘ answered’ by science. Society is going to have to decide who has access to our genetic profile. For example, should we all be on a DNA database to help the police to catch criminals? Or is this a step too far, because in the future it could allow employers and insurance companies access to data we would rather they did not see? Changing values Scientific data does not change according to who is reading it. But values can change, especially in different environmental and social contexts. For instance, many Roman Catholics think abortion is a sin, but many other people do not and decide to have an abortion because they think it is the right thing to do. Cloning occurs naturally in some plants and in human identical twins, and it is now possible to clone animals artificially. The subject of cloning raises a number of issues which need to be taken into consideration. Clones Clones

are genetically identical individuals. Bacteria, plants, and some animals, can reproduce asexually to form clones that are genetically identical to their parent. Identical human twins are also clones. Any differences between them are due to environmental factors. Asexual reproduction only requires one parent, unlike sexual reproduction, which needs two. Since there is only one parent, there is no fusion of gametes, and no mixing of genetic information. As a result, the offspring are genetically identical to the parent, and to each other. They are clones. Plants Asexual reproduction in plants can take a number of forms. Many plants developunderground food-storage organs that later develop into the following year’s plants. Potato plants and daffodil plants do this. Daffodil bulb at start of season Daffodil bulb at end of season Strawberry plant with runners - stems growing sideways Some plants produce side branches withplantlets on them. The Busy Lizzie plant does this. Others, such as strawberry plants, produce runners with plantlets on them. Animals Asexual reproduction in animals is less common than sexual reproduction. It happens in sea anemones and starfish, for example. Natural cloning Twins are genetically identical because they are formed after one fertilised egg cell splits into two cells. They are natural clones. Artificial cloning It is possible to make clones artificially. The cloning of animals has many important commercial implications. It allows an individual animal that has desirable features, such as a cow that produces a lot of milk, to be duplicated several times. Stem cells During the development of an embryo, most of the cells become specialised. They cannot later change to become a different type of cell. But embryos contain a special type of cell called stem cells. These can grow into any type of cell found in the body. They are not

specialised. Stem cells can be removed from human embryos that are a few days old, for example, from unused embryos left over from fertility treatment. Here are some of the things stem cells could be used for: * making new brain cells to treat people with Parkinson’s disease * rebuilding bones and cartilage * repairing damaged immune systems * making replacement heart valves Therapeutic cloning If you were to receive medical treatment with cells grown from stem cells, your body’s immune system would recognise the cells as foreign, and they would be rejected and die. But this would not happen if you received cells with the same genes as you. This could be done by cloning one of your cells to produce an embryo, then taking stem cells from this. This is called therapeutic cloning. Here are the steps involved: 1. nucleus taken out of a human egg cell 2. nucleus from a patient's cell put into the egg cell 3. egg cell stimulated to develop into an embryo 4. stem cells taken from the embryo 5. stem cells grown in a container of warm nutrients 6. stem cells treated to develop into required cell types Ideas about science - weighing up arguments The cloning of human embryos is a very contentious issue. You need to be able to use your Ideas about Science to: * Say clearly what the issues are. For example, some human embryos will be destroyed during the cloning process. * Summarise different views that might be held. For instance, some people think that this is murder. Others believe it is furthering our knowledge of science for the benefit of everyone. * Identify and develop arguments based on the idea that the right decision is the one that has the best outcome for the majority of people. For example, even though a few embryos will be destroyed, many people will be free from suffering and cured of diseases that are currently

incurable. * Identify and develop arguments based on the idea that certain actions can never be justified because they are unnatural and wrong. For example, some people think that, no matter what the benefits are of curing a disease, we should not clone embryos because destroying human life is always wrong. Remember that these types of decisions are values, and produce questions that cannot be answered by science. Artificial cloning of animals — higher There are two main ways to clone animals. Embryo transplants A developing embryo is removed from a pregnant animal at an early stage, before its cells have had time to become specialised. The cells are separated, grown for a while in a laboratory, then transplanted into host mothers. When the offspring are born, they are identical to each other, and to the original pregnant animal. They are not identical to their host mothers, because they contain different genetic information. Fusion cell cloning Fusion cell cloning involves replacing the nucleus of an unfertilised egg with one from a different cell. The replacement can come from an embryo. If it is from an adult cell, it is called adult cell cloning. 'Dolly the sheep' was the first mammal to be cloned using adult cell cloning. She was born in the UK in 1996, and died in 2003. Here is how she was produced: 1. An egg cell was removed from the ovary of an adult female sheep, and its nucleus removed. 2. The nucleus from an udder cell of a donor sheep was inserted into the empty egg cell. 3. The fused cell then began to develop normally, using genetic information from the donated DNA. 4. Before the dividing cells became specialised, the embryo was implanted into the uterus of a foster mother sheep. The result was Dolly, who was genetically identical to the donor sheep. The cloning process of 'Dolly the sheep'