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TEACHER'S NOTES
EXPLORING THE RISKS

OVERVIEW **Aimed at key stage 4 pupils. This is a worksheet activity to help pupils understand how Huntington's disease (HD) is inherited and the risks involved.**

LEARNING OBJECTIVES

- To understand how HD can be passed from parent to child
- To appreciate that some people use pre-implantation genetic diagnosis (PGD) to avoid passing the condition on

CURRICULUM LINKS

- **KS4:** The ways in which organisms function are related to the genes in their cells
- **KS4:** Human health is affected by a range of environmental and inherited factors, by the use and misuse of drugs and by medical treatments

Activity

- Introduce the genetic condition - Huntington's disease
- Show the film Luke's story on **www.genesareus.org**
- It would be helpful to complete the worksheets on 'Family trees' and 'Testing embryos' before using this worksheet
- Give pupils the worksheet and ask them to complete questions 1 – 6
- Questions 7 & 8 are discussion questions, which could be explored in small groups or with the whole class

ANSWERS

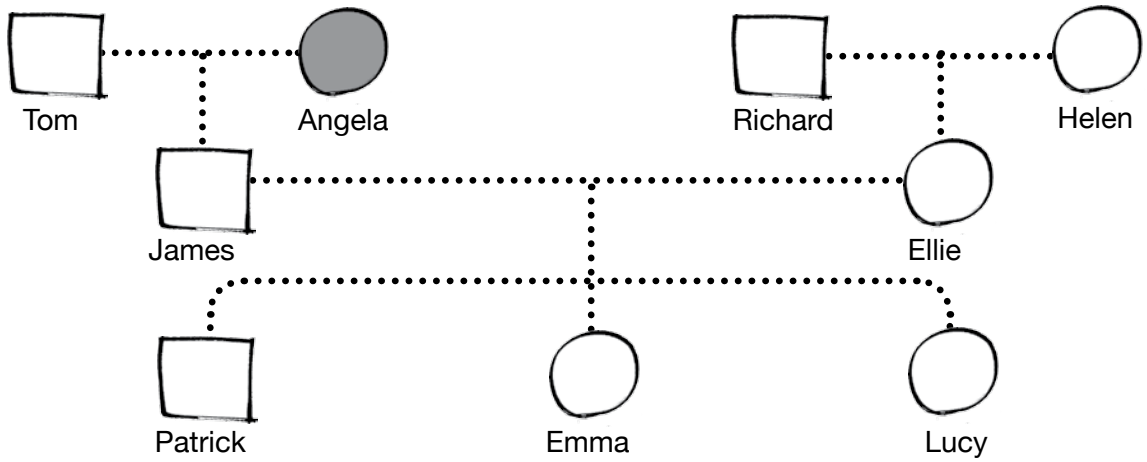
1. Explain the difference between a dominant and recessive allele

The effects of a dominant allele can be seen in someone's phenotype when someone only has one copy of the allele. When an allele is recessive, someone needs to have two copies of the allele in order to see the effect on their phenotype.

2. Approximately how many people have HD in the UK? (base your calculations on there being 60 million people living in the UK)

If 1 person in 10,000 is affected by HD, then we would expect 6000 people to be affected across the UK.

3. Draw out a family tree (also called a 'pedigree') based on the following information



4. Using the family tree you just drew, answer the following questions:

a. What is the probability that James has inherited the HD-causing allele?

50% or 0.5

b. What is the probability that Ellie has inherited the HD-causing allele?

Zero - though it is worth being aware that the mutation that causes HD can arise spontaneously, so there is technically a chance that Ellie has inherited a *de novo* mutation, but this would be incredibly unlikely.

c. Without knowing whether James has the HD-causing allele, what is the probability that Patrick has inherited the HD-causing allele?

25% or 0.25

ANSWERS continued

d. If James discovers he has the HD-causing allele, how does that affect the probability that Patrick has inherited this allele?

Patrick's risk rises to 50% or 0.5

e. If James discovers he does not have the HD-causing allele, how does that affect the probability that Patrick has inherited it?

Patrick's risk falls to zero (with the same caveat as in 'b')

5. James and Patrick discover they have the HD-causing allele; how does that affect the probability of Emma and Lucy also having the allele?

It does not affect their probability – the likelihood of James passing on the HD-causing allele to any of his children is 50% for each child, regardless of his other children.

6. Patrick marries Sarah; how does that affect Sarah's likelihood of developing HD?

This does not affect Sarah's risk of developing HD. She is at the population risk (in other words - she is incredibly unlikely to develop HD).

7. If you knew you had a 50% probability of passing on a serious condition like HD to your children, would that affect how you would feel about having children?

There is no right or wrong answer to this. Pupils should try to describe how they would feel in this situation. They might describe not wanting to have any children or using genetic testing in pregnancy to avoid passing the condition on. They might feel that they would want to find out if they had the HD-causing allele before deciding to have children or they might decide to have children and leave the outcome 'in the lap of the gods'.

8. Why do you think PGD is considered controversial, and in what instances do you think it is used?

PGD is sometimes considered controversial because healthy embryos can be discarded as part of the process and some would argue that it borders on eugenics. It is worth noting that healthy embryos are also discarded during routine IVF procedures. At present PGD is limited to checking for specific conditions, as outlined by the HFEA (see links below) and only couples at risk of passing on a particular condition to their children are allowed to undergo PGD.

FURTHER information

- The Huntington's Disease Association (HDA) provides excellent information about the condition: **http://hda.org.uk**.
- American website called 'Your Genes, Your Health' has animations and films to help explain HD for school pupils. It is pitched above GCSE-standard, but would be accessible to able pupils: **www.ygyh.org**.
- The Human Fertilisation and Embryology Authority (HFEA) website has excellent information introducing PGD and explaining what genetic conditions can be tested for **www.hfea.gov.uk**.

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STUDENT WORKSHEET

EXPLORING THE RISKS

Huntington's disease (HD) is a genetic condition.That means that somebody with HD has inherited it, via their genes, from one of their parents. Roughly 1 person in every 10,000 is affected by HD in the UK. Because HD is genetic it can only be inherited from our parents; HD is not contagious.

We inherit two copies of every gene – one comes from our mum, the other from our dad. Quite often the two copies will be slightly different from each other. When there are two different versions of the same gene they are called 'alleles'. Usually the slight differences between the two copies of the same gene do not affect our health. However, sometimes a change in one of the copies can have detrimental effects on our health and cause a genetic condition, like HD. When having just one version of an allele is enough to affect a person, that allele is described as dominant.

The allele that causes HD is dominant and people affected by HD nearly always have one 'healthy' allele and one changed allele. It is very rare for somebody with HD to inherit two copies of the HD-causing allele. If somebody who has an HD-causing allele has children with somebody who has only 'healthy' alleles there is a 50% chance that any child will inherit the HD-causing allele. The risk of the HD-causing allele being passed on in this situation is often compared to flipping a coin (it's a 50:50 risk). The sex of the child does not alter the risk of inheriting this condition. Some genetic conditions are sex-linked, but this only happens when the gene involved is located on the X or Y chromosome.

1. ***Explain the difference between a dominant and recessive allele***

2. ***Approximately how many people have HD in the UK? (base your calculations on there being 60 million people living in the UK)***

3. ***Draw out a family tree (also called a 'pedigree') based on the following information:***
   - Patrick, Emma and Lucy are siblings
   - James and Ellie are the parents of Patrick, Emma and Lucy
   - Richard and Helen have a daughter called Ellie
   - Tom and Angela have a son called James
   - Angela has HD

4. ***Using the family tree you just drew, answer the following questions:***
   - ***a. What is the probability that James has inherited the HD-causing allele?***
   - ***b. What is the probability that Ellie has inherited the HD-causing allele?***
   - ***c. Without knowing whether James has the HD-causing allele, what is the probability that Patrick has inherited the HD-causing allele?***
   - ***d. If James discovers he has the HD-causing allele, how does that affect the probability that Patrick has inherited this allele?***
   - ***e. If James discovers he does not have the HD-causing allele, how does that affect the probability that Patrick has inherited it?***

5. ***James and Patrick discover they have the HD-causing allele; how does that affect the probability of Emma and Lucy also having the allele?***

6. ***Patrick marries Sarah; how does that affect Sarah's likelihood of developing HD?***

7. ***If you knew you had a 50% probability of passing on a serious condition like HD to your children, would that affect how you would feel about having children?***

Pre-implantation genetic diagnosis (PGD) is a way of finding out whether an embryo conceived by *in vitro* fertilisation (IVF) has any genetic changes that could lead to health problems. PGD is sometimes used to test embryos for the HD-causing allele. This type of genetic testing is quite controversial and PGD is not done very often (about 100 babies are born each year after PGD in the UK).

8. ***Why do you think PGD is considered controversial, and in what instances do you think it is used?***