

UNIT 6: WEIGHING THE PROS AND CONS

TEACHERS' NOTES UNIT 6

WEIGHING THE PROS AND CONS

OBJECTIVES

To encourage students to consider the benefits and disadvantages to the consumer of being screened/tested for different genetic or chromosomal conditions.

SUGGESTED AGE RANGE

14-16 year olds

CURRICULUM LINKS

For use in PSE, Religious and Moral Education, Social and Contemporary Studies, English etc.

SUMMARY OF CONTENT

Students work in pairs through one of three Case Studies which use real-life examples to explore some of the implications for individuals and families of screening for different genetic and chromosomal conditions (Tay Sachs Disease, Genetic Breast Cancer and Down's Syndrome). Students are then presented with a magazine advertisement which offers genetic testing through the post, and asked to think about whether or not they would like to be screened to see if they are carriers of the Cystic Fibrosis gene.

TEACHERS PLEASE NOTE

You may like to read the Notes for Teachers on the Case Studies on pages 67—69 and Notes for Teachers on Activity Sheet A on page 70 before the lesson. It is important to be aware of the fact that some students in the class may themselves have a genetic condition, or be a carrier, or have relative who is affected. Sensitivity is required to avoid putting such students under stress

MATERIALS NEEDED

Photocopies of whichever Case Studies you have selected for use with your students (there are three to choose from), plus the Genetic/ Chromosomal Condition Card which goes with each Case Study:

Case Study IA (Daniel) plus Genetic Condition Card: Tay Sachs Disease

Case Study IB (Lynne) plus Genetic Condition Card: Genetic Breast Cancer

Case Study IC (Monica and Sami) plus Chromosomal Condition Card: Down's Syndrome

One Case Study and Genetic/Chromosomal Condition Card per pair of students is required.

One copy of Activity Sheet A plus Genetic Condition Card: Cystic Fibrosis per pair of students.

WHAT YOU DO

Explain to students that scientific and medical knowledge about genes is increasing all the time. One result is that tests have been, and are being, developed which will tell people if they have a faulty copy of a particular gene which could:

- Be passed onto the next generation
- Cause disability and/or disease.

This section asks students to think about some of the possible consequences of such tests.

Ask students to work in pairs. Give each pair a copy of the Case Study plus Genetic/Chromosomal Condition Card you have selected for them and ask students to work through the questions on the Case Study in their pairs. Set a time limit for the activity before coming back together as a whole class to process the activity (you can use the Notes for Teachers on the Case Studies to help you. The Glossary is also available to help with words which might be unfamiliar either to you or to students e.g. carrier, testing, screening etc.

TEACHERS' NOTES UNIT 6

WHAT YOU DO

Then hand out copies of Activity Sheet A plus Genetic Condition Card: Cystic Fibrosis and either work through the sheet as a whole class or again in pairs. You can use the Notes for Teachers on Activity Sheet A on page 70 to help you process the activity.

NOTES ON CASE STUDY IA — DANIEL

1. Daniel's mother is a carrier but his father is not, which means that Daniel has a 50% chance of being a carrier of the Tay Sachs Disease gene. If Daniel has the test and it shows he is a carrier, he will have a number of choices if/when he decides to have children:
 - a) say nothing and hope his partner is not a carrier
 - b) explain to his partner that as he is a carrier, it might be a good idea for her to be tested to see if she is also a carrier. If she is, each and every child of theirs would have a 25% (1 in 4) chance of having Tay Sachs Disease, a 50% (1 in 2) chance of being an unaffected carrier like themselves, and a 25% (1 in 4) chance of neither having Tay Sachs Disease nor being a carrier. Genetic counselling should help them to think about their options and what they want to do. If Daniel does not take the test, he will not know whether or not his children are at risk from Tay Sachs, and his choices are more limited.
2. There is no 'best time' for an adult to be tested. Informed consent is important, as is counselling to make sure that the person understands the implications of the test and what their options are when they get the results. Screening for carrier status before starting a family means that the person can make a more informed reproductive choice.
3. Daniel doesn't have to tell anyone that he is a Tay Sachs carrier. But it would probably be wise to tell his partner before they decide to have children (if she is an Ashkenazi Jew, she has a higher than average chance - 1 in 25 as opposed to 1 in 250 of the general population - of being a carrier of the Tay Sachs Disease gene).

A few people who have tested positive for carrier status for a genetic condition have subsequently found that they have been discriminated against. This is because some employers, insurance companies and other institutions have wrongly assumed that being a carrier is the same as having the disease.

NOTES ON CASE STUDY 1B — LYNNE

N.B. Genetic Breast Cancer is usually known as 'familial breast cancer'. We have used the term 'genetic' in order to remind students that this form of the disease is caused by a faulty gene.

When Lynne is being counselled to help her decide whether or not she wants to have the test, the following points are likely to be discussed:

- If Lynne doesn't have the test, will she spend a lot of time and energy worrying about getting Breast Cancer, and about whether or not her two daughters also have the faulty gene?
- If Lynne does have the test, and it shows that she does not have a faulty copy of the BRCA1 gene, she need not worry about either herself or her daughters being any more at risk of developing Breast Cancer than the rest of the population. The relief of knowing this may be tempered by feelings of guilt and sadness as so many of her relatives are affected.
- If Lynne has the test and it shows that she does have a faulty copy of the BRCA1 gene, she will have to live with knowing that she is likely to develop Breast Cancer, possibly while she is in her 30s or 40s, and that she might also have passed the faulty gene onto her daughters.
- A few women who have been tested and know that they have a faulty copy of the BRCA1 gene have chosen to have a preventative double mastectomy while they are showing no signs of cancer. They may be offered full mastectomy (the surgical removal of both breasts) or subcutaneous mastectomy (where as much breast tissue as possible is removed from both breasts and replaced with an implant). At present, there is no hard scientific data on how effective the two methods are at preventing Breast Cancer.
- There is no 'best time' for Lynne to be tested. The most important thing is that she has counselling to help her to fully understand her options and to reach a decision which is right for her (and her family). If she is thinking in terms of prevention rather than cure, she will want to consider testing for the gene before she develops Breast Cancer.

TEACHERS' NOTES UNIT 6

Genetic conditions have implications not just for affected individuals but for whole families

Telling other family members about a diagnosis can be difficult even when relationships within a family are good, but Lynne's genetic counsellor might be able to help Lynne by talking through whether or not, and how, she could tell her sister.

NOTES ON CASE STUDY IC — MONICA AND SAMI

It is important for all women who have fetal screening during pregnancy to understand what each test can (and cannot) tell them about their developing fetus. It is also important that these women have an opportunity to think and talk about what they will do if the test reveals some form of fetal abnormality.

In some health authorities, older women (i.e. aged 35 and above, although this can vary) are routinely offered amniocentesis during pregnancy. A woman who has amniocentesis while she is pregnant should be aware of the following:

The test appears to cause about 1 in 100 women to lose their baby through miscarriage.

The test tells a woman whether or not her fetus has Down's Syndrome. It does not guarantee a 'perfect' baby. Having the test and waiting for the results can cause anxiety in some women. A few women continue to worry even when they have been told that the fetus is at low risk of having Down's Syndrome.

Any woman who has the test needs to think in advance about what she will do if she is told that her fetus has Down's Syndrome. Will she terminate the pregnancy? What information and support is available to help her reach a decision? Should women refuse the test unless they are willing to consider abortion? Or is it helpful for all older women to have the test so that if the fetus is affected, they can choose between abortion or continuing with the pregnancy?

This is a matter of opinion. Abortion is a legally available option in this country. Many people support the right of women to decide whether or not to continue with an affected pregnancy as they are the ones who will have to live with the consequences of that decision, and to be denied that choice is felt to be discriminatory. Others would prefer that choice not to be available and believe that abortion is unacceptable in all circumstances.

Some disabled people argue that attempting to eradicate congenital disability through prenatal fetal screening is a form of discrimination because it implies that a woman who finds out that her fetus is impaired will terminate the pregnancy. They point out that disability will never be eradicated through prenatal screening as most impairment occurs through disease, the ageing process, accidents etc., and that reducing the number of babies born with some form of disability will simply lead to further isolation and prejudice against disabled people. However, the experience of the majority of parents who have a child with a genetic disorder and who use selective termination to avoid the birth of an affected sibling is that they love the child with the disorder no less as a result of the choices they have made. Indeed in the case of serious genetic conditions, the choice to terminate may be motivated in part by a desire to be able to offer the affected child the time and resources he/she needs.

The advertisement similar to the one featured in Activity Sheet A appeared in the classified section of *Cosmopolitan* in August 1995. It is offering a test to detect carriers of the Cystic Fibrosis gene, and is aimed at women who are planning pregnancy and who are prepared to pay for personal genetic information. The test costs approximately £65 per person or £100 for a couple. Around 1 in 25 of the UK population carries one faulty copy of the gene but you have to have two faulty copies to be affected by the disease.

WHAT IS CARRIER SCREENING?

Genes work in pairs, one copy in each pair coming from our mother and one copy from our father. Cystic Fibrosis is an example of a recessively inherited genetic condition, which means that a person has to inherit two faulty copies of the gene (one from each parent) to be affected by Cystic Fibrosis. A person who inherits one working copy of the gene and one faulty copy will not develop Cystic Fibrosis because

Carrier Screening continued

the working copy functions normally and masks the effects of the faulty gene. That person will, however, be a carrier and be able to pass the faulty gene onto any children they may have.

Carrier screening may be offered to people not known to be at risk to see if they carry a copy of the faulty gene. The purpose of this type of screening is to help people make informed decisions regarding reproduction. For example, if you find out that you are a carrier of a faulty gene for Cystic Fibrosis and you want to have children, you know that each and every child of yours has a 50% (1 in 2) chance of inheriting the faulty gene and being an unaffected carrier like yourself.

TEACHERS' NOTES UNIT 6

But if your partner also carries a faulty copy of the same gene, each and every child of yours also has a 25% (1 in 4) chance of having Cystic Fibrosis, and a 25% (1 in 4) chance of being neither a carrier nor having the condition.

GENERAL POPULATION SCREENING

Some of the factors which need to be taken into account when setting up a screening programme for carrier status within the general population include:

- Does a reliable test exist?
- Is the condition common and important/serious?
- Is screening cost effective?
- Do the benefits of having this information outweigh any potential disadvantages?
- When should this screening be offered? (i.e. at birth, to older children, to individuals or couples before pregnancy, or during pregnancy?)

As far as Cystic Fibrosis carrier screening is concerned, a test is available which can accurately detect up to 80% (but not all) of the mutations in the gene, so there is a chance that you might be a carrier of one of the less common mutations and the test would not show this. Being a carrier does not mean that you will ever develop Cystic Fibrosis, but if you had a child with another carrier, that child could have Cystic Fibrosis, a serious genetic condition which (given the choice) no-one would want their child to have. Some people say that knowing one's carrier status allows one to make appropriate reproductive decisions, others that it is an infringement of one's right not to know and can lead to discrimination by employers and/or insurance companies who do not understand the difference between being a carrier and being affected by a genetic disorder.

COMMERCIAL SCREENING

Commercial screening by post is now available to anyone willing to pay to find out if they are an unaffected carrier of the Cystic Fibrosis gene. However, there are concerns about the regulation of such tests, and the fact that face-to-face counselling to help people interpret and cope with the results is not routinely offered by commercial testing companies. This has led to fears that people will simply ignore or fail to understand the reproductive implications of a positive test result, or that having paid for a private test, they will then need to approach the health service for explanations and reassurance and perhaps for a repeat test to check the validity of the original results. The Advisory Committee on Genetic Testing (ACGT) has drawn up a code of practice in order to try and overcome some of these difficulties and to establish requirements - especially in terms of efficacy and product information — to be met by commercial suppliers of genetic tests.

CASE STUDIES FOR STUDENTS UNIT 6

WEIGHING THE PROS AND CONS



CASE STUDY IA

DANIEL

Anna is 45. She knows that she is a carrier of a faulty copy of the Tay Sachs Disease gene. She has never really talked to her son Daniel about the possibility that he might also be a carrier. After all, being a carrier doesn't make you ill — it's not the same as having Tay Sachs Disease. In fact you probably wouldn't even know you were a carrier unless you'd been tested.

But now Daniel is 18 and has a girlfriend. One day perhaps he will want to have children. Supposing he married a woman from the Jewish community like himself and they were both carriers of a faulty copy of the Tay Sachs Disease gene? Any baby of theirs would have a 25% (1 in 4) chance of being born with Tay Sachs Disease and of dying while still very young.

Anna is not sure if Daniel knows all this. So one day she suggests that he should think about being tested to see if he is a carrier.

TASK

- a) Read the Genetic Condition Card on Tay Sachs Disease.
- b) Fill in your answers to the questions in the box below.

1. If you were Daniel, would you have the test?

YES/NO because

2. If Daniel decides to have the test and finds that he is a Tay Sachs carrier, who do you think he should tell, and why:

- a) His mother?
- b) His girlfriend?
- c) Any woman he has a sexual relationship with?
- d) Any woman he wants to have children with?
- e) No-one?

Because...

CASE STUDIES FOR STUDENTS UNIT 6

CASE STUDY 1B

LYNNE

Lynne is 30 and has two daughters aged three and 18 months. She knows that 15 female relatives on her father's side of the family have developed Breast Cancer and is beginning to worry that she too might be at risk of getting the disease. She has heard something about a Breast Cancer gene and decides to visit her doctor to find out more.

Lynne is told that the fact that so many members of her family have developed Breast Cancer suggests that there is a 50% chance that she has inherited a faulty gene, which can cause Breast Cancer. If she has, there is an 80—90% chance that she will develop Breast Cancer. She could also have passed a copy of the faulty gene on to her daughters. Her doctor suggests that she should think about being tested to see if she has inherited the faulty gene and refers her to her Regional Genetics Centre. Only women from families where there is a high incidence of Breast Cancer are offered this test.

Lynne has special counselling to help her decide what to do. If she has the test and it shows that she has inherited a faulty copy of the gene, she knows she is likely to develop Breast Cancer in her 30s or 40s. But if she has the test and it shows that she does not have the faulty gene, her Breast Cancer risk is no higher than that of most other women. And if she doesn't have the faulty gene herself, she can't have passed it on to her daughters either.

TASK

- a) Read the Genetic Condition Card on Genetic Breast Cancer
- b) Fill in your answers to the questions in the box below.

1. Do you think Lynne should have the test?

YES/NO because

2. When do you think would be the best time for Lynne to have the test, and why:

- a) As soon as possible?
- b) When her daughters are older?
- c) Around the time she is most likely to develop Breast Cancer?
- d) Before she has any more children?
- e) Never because...

CASE STUDIES FOR STUDENTS UNIT 6

3. Lynne has a sister living abroad. If Lynne has inherited a faulty copy of the gene from her father, it is possible that Lynne's sister has inherited a faulty copy of the gene from him too. Lynne and her sister don't get on and have very little contact. If Lynne has a test and finds that she has a faulty copy of the gene, do you think she should tell her sister?

YES/NO because...

CASE STUDIES FOR STUDENTS UNIT 6

CASE STUDY 1C

MONICA AND SAMI

Monica and Sami have two adopted children aged nine and seven. Now, at the age of 38, Monica is pregnant for the first time and the whole family are delighted.

Monica's doctor is suggesting that as she is 38 years old, she should have a test called amniocentesis (pronounced am-nee-yo-sen-tea-sis) when she is about 16 weeks pregnant.

Amniocentesis involves drawing out a small amount of the fluid which surrounds the fetus and examining cells in this fluid. The test can accurately predict if the fetus is affected by Down's Syndrome. Monica is being offered this test because older mothers (aged 35 and over) have a greater chance of having a baby with Down's Syndrome. A few women (about 1 in every 100) will lose their unborn baby through miscarriage because of the test.

Monica is worried about the test. This pregnancy is very precious to both Monica and Sami as Monica was told years ago that she was infertile and would never be able to have a baby.

TASK

- a) Read the Chromosomal Condition Card on Down's Syndrome.
- b) Fill in your answers to the questions in the box below.

1. Do you think Monica should have the test?

YES/NO because...

2. What do you think Monica and Sami should do if the test shows that their unborn baby has Down's Syndrome?

I/we think...

3. Do you think it is important that women are allowed to choose whether or not to terminate an affected pregnancy, or do you think that abortion is always wrong?

I/we think...

CASE STUDIES FOR STUDENTS UNIT 6

4. Some people say that aborting a fetus on the grounds of disability is a form of discrimination against disabled people. What do you think?

I/we think...

ACTIVITY SHEETS FOR STUDENTS UNIT 6

ACTIVITY SHEET A

The advertisement similar to the one below appeared in the health and beauty section of a popular magazine. Read through the advertisement and the Genetic Condition Card on Cystic Fibrosis. Then work through the questions for discussion.

PLANNING PREGNANCY?
Cystic Fibrosis
—AND YOUR FUTURE CHILDREN—

Two million people in the UK carry the Cystic Fibrosis gene and show no outward sign of it - you could be one of them.

If you are a carrier you will not suffer from this chronic disorder of the lungs and digestive system, but the quirks of inheritance mean that your future child might if your partner is a carrier. Nearly every day one affected child is born to parents who did not know that they were carriers of the gene, would you?

The NHS does not routinely offer CF carrier screening but an established, effective and inexpensive test is available.

A simple mouthwash sample, taken in the privacy of your home, is all that we require to determine whether you are carrying this disorder. You will receive your results and a full explanation within days. A Clinical Genetics Consultant is also available to provide helpful information and advice at no extra fee.

QUESTIONS FOR DISCUSSION

- What is this advertisement for?
- Men and women can carry a faulty Cystic Fibrosis gene without knowing it, so why do you think the company offering this test decided to advertise in a magazine aimed mainly at young women?
- Screening the general population to see if they carry a faulty Cystic Fibrosis gene is not a routine part of health care. Should it be? Do you think doctors ought to screen everyone free of charge?
- What are some of the advantages and disadvantages of the type of screening offered in the advertisement (i.e. where you send off your mouthwash sample in the post and the company writes back to you with the results)?
- Would you agree to be screened for the Cystic Fibrosis gene? Why/why not?

Please send me the free information leaflet on Cystic Fibrosis and the gene test