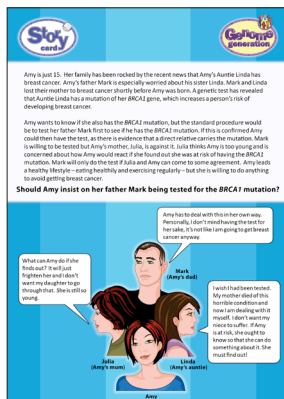


GENOME GENERATION

Guide to scenario 1

AMY AND THE BREAST CANCER TEST



This scenario is set in the present day.

Summary: Scenario 1 deals with a 15 year old girl (Amy) who has a family history of breast cancer. Her paternal aunt has been diagnosed with the disease and tests have shown that she is a carrier of the *BRCA1* mutation associated with breast cancer. There is a dispute within the family about whether Amy's father should be tested to find out whether he is also a carrier of the *BRCA1* mutation, and may have passed the risk to Amy. Her mother does not want the test to take place. The decision is Amy's. The group has to decide whether Amy should ask her father to take the test.

Initial question: Should Amy insist on her father being tested for the *BRCA1* mutation?

Key issues: Would you want to know? Impact on families

SENSITIVE ISSUES & GUIDANCE

This scenario is appropriate for GCSE and A-level.

The issues explored deal with the impact on families, and the question of whether or not you would want to know, if you were faced with a predisposition to a genetic condition.

The fact that the main character is young might make it easier for students to relate to the scenario. Be aware that some people in the group may have a family history of breast cancer.

QUESTIONS TO DISCUSS

Amy is only 15 years old. Is she too young to deal with the information or will it give her an opportunity to change her lifestyle accordingly?

An awareness of her carrier status could mean that Amy is monitored continuously and that any sign of the disease is spotted and eradicated early.

What impact might the test have on Amy in later life with regards to the decisions she makes?

If Amy discovers that she is a carrier of the *BRCA1* mutation, should she increase her chances of survival with a pre-emptive mastectomy, or is she too young to take such drastic action (for a condition that she may never get)?

Can men really get breast cancer?

Amy's father Mark might not realise that he is also at risk. His character quote implies that he wrongly assumes that men cannot get breast cancer.

A negative *BRCA1* test for Mark does not mean Amy will definitely not develop the disease. She may develop breast cancer regardless of whether she has the *BRCA1* mutation. Would the false confidence that she is 'in the clear' lead her to be complacent?

Why is Mark (the father) being tested rather than Amy?

This is because in current clinical practice, it is unlikely that a 15 year old would be tested. They would first test the Aunt to discover the exact mutation they are looking for. The parent (in this case, her father) would then be tested to see if they also carry the mutation. This may raise a variety of questions about consent. Should there be a minimum age of consent for genotyping to take place? Should Mark allow his daughter and wife access to his private data?

FURTHER INFORMATION – WHAT THEY MIGHT NEED TO KNOW

What are *BRCA1* and *BRCA2* genes?

BRCA1 and *BRCA2* are tumour suppressor genes – their role is to prevent cells from becoming cancerous. Mutations in the *BRCA1* and *BRCA2* genes can affect the cell's ability to repair DNA. If repair is inefficient, mutations in DNA can persist and result in cancer. Many variations of *BRCA1* and *BRCA2* genes have been found, some of which are associated with an increased risk of breast cancer. Mutations in *BRCA1* also increase the risk of ovarian, fallopian tube and prostate cancer.

How accurate are genetic tests for *BRCA* mutations?

No test is 100% accurate. Genetic tests can generate false negatives and false positives. However, tests for *BRCA* mutations are very reliable. They usually pick up 98% of mutations, with less than 1% being false positives or negatives. Once a *BRCA* mutation has been identified in an affected family member, finding mutations in other family members is easier because clinical geneticists know exactly what to look for. However a negative test does not necessarily mean that a person is not at risk, even if it is not a false negative. Not all breast cancer cases are due to a mutation in the *BRCA* genes. Only about 3% of cases of breast cancer are related to a known inherited breast cancer gene.

Other genes and breast cancer

BRCA1 and *BRCA2* are not the only genes associated with an increased risk of breast cancer. Other genes which have been identified as important are *TP53* and *PTEN*. These are also tumour suppressor genes and mutations in these genes are frequently involved in the development of cancer. Mutations in 10 additional genes have also been associated with increased risk of breast cancer but as yet, there are no clinical tests for these.

Genetic screening of cancerous cells to determine appropriate treatment

Cancer cells carry mutations that make them genetically different from other cells. The specific genetics of the cancerous cells can be important in selecting the best treatment. As we increase our understanding of genetic differences (and how our genes affect response to cancer therapies) we are able to improve treatments and target them more accurately. Examples of anti-cancer drugs that target particular genetic mutations are trastuzumab (Herceptin) and tamoxifen.

Risk factors for breast cancer

Established risk factors for breast cancer (in addition to being female) include, late or no childbearing, breastfeeding, early onset of periods, late menopause, use of the pill, use of HRT (hormone replacement therapy), high breast density, family history of breast cancer, being overweight or obese, lack of physical activity and high alcohol consumption. Rates of breast cancer are increasing in the UK.

FURTHER READING

The following news stories can be used in addition to the Info cards to provide extra background information to help students understand the major issues raised in this scenario.

Should I have the breast cancer gene test?

www.bbc.co.uk/news/health-14135296

Paternal health 'disregarded in breast cancer cases'

www.bbc.co.uk/news/health-11607396

Gene dilemma to prevent next generation cancer

www.bbc.co.uk/news/health-14012561

FURTHER INFORMATION FOR STUDENTS

Students can find out more information about breast cancer from the following websites:

NHS Choices

NHS Choices is produced by the National Health Service (NHS) and provides information on a range of different health issues. The pages below provide information on breast cancer.

www.nhs.uk/Conditions/Cancer-of-the-breast-female/Pages/Introduction.aspx

www.nhs.uk/conditions/cancer-of-the-breast-male/Pages/Introduction.aspx

GENOME GENERATION

Guide to scenario 1

Breast Cancer Care

Breast Cancer Care is a UK charity that provides information and support for anyone affected by breast cancer.

www.breastcancercare.org.uk

Cancer Research UK

Cancer Research UK is the world's leading cancer charity. It provides funding for research and provides information on cancer for patients, healthcare professionals and the public.

www.cancerresearchuk.org/about-cancer/type/breast-cancer/

FOLLOW UP ACTIVITY

Students will take the role of the genetic counsellor at the hospital where Mark, Amy's father, will be getting his *BRCA1* genetic test. They have to create a pedigree diagram based on the information provided for him. All information to complete this task is provided in accompanying worksheets (see Scenario 1 follow up activity).