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Cancer in the Family

To be read in conjunction with reference sheets C and D

Chloe, who is 21, comes to a genetics clinic to discuss with a cancer geneticist her family history of breast cancer. Her paternal uncle, Bruce, was diagnosed with breast cancer in 1999 at the age of 48.

Two genes associated with an increased risk of breast cancer were identified in 1994. The BRCA1 gene on chromosome 17q21, and the BRCA2 gene on chromosome 13q12.3.

Having a mutation in BRCA1 or BRCA2 does not mean you will definitely get breast cancer. It means you have a predisposition for developing breast cancer and are at a much greater risk than the general population.

The BRCA genes only explain 1 or 2% of all breast cancer.

About 1 in 1000 people carry a BRCA1 or BRCA2 gene mutation.

Chloe's uncle, Bruce, attended a familial cancer clinic to have genetic testing and was found to have a mutation in his BRCA2 gene. Chloe's father, Wayne, has another daughter, Michelle, from a previous relationship. Michelle is 24 years old but is estranged from her father. Chloe also has a younger brother, Matthew.

1. What is the chance that Wayne could also have the BRCA2 mutation?

Chloe wants to find out whether she too is a carrier of a BRCA2 mutation.

http://www.accv.org.au
http://www.nbcc.org.au

For all Australian women, the lifetime risk of developing breast cancer is 1 in 13.

Her father, Wayne, refuses to find out his genetic status and is unwilling to discuss testing or the family's history of cancer. Gene testing is offered to Chloe, based on Bruce's test results, and she decides to undertake the test after discussing it fully with the cancer geneticist and a genetic counsellor.

2. What is Chloe's risk of having inherited the BRCA2 gene from her father who has not undertaken gene testing?

3. What advantages can you see for Chloe in knowing her genetic status for the BRCA2 gene?

4. Wayne is reluctant to have a gene test. Suggest two reasons why he may see this information as disadvantageous.

5. Describe two benefits for Wayne of having the gene test.

Chloe thinks that it is important that her half-sister, Michelle, is told that a breast cancer gene mutation has been found in the family. Michelle lives in Perth and has had no contact with Wayne in recent years.

6. What do you think Chloe should do about Michelle? How should she go about it?

7. Do you think that gene testing for the family mutation could be an issue for Chloe's brother, Matthew, when he is older? Why?