It's all in your genes Can our genes tell us to how to treat and prevent disease?

Sue's story

Sue is a 48 year-old woman who has two children: a 17 year-old boy and a 22 year-old girl. She has always led a healthy and active lifestyle. One morning when showering, she noticed a lump in her breast. She went straight to her GP who referred her to the breast clinic at the hospital where she was examined and went for X-rays and had a biopsy (small tissue sample) of the lump taken.

The results of the biopsy confirmed that Sue had breast cancer. The tumour was staged at T2N1M0. This means that the tumour was 2-5cm across (T2); that it had spread to the lymph glands in her armpit (N1); but showed no signs of spread to any other part of her body (M0).

Like all newly diagnosed patients with breast cancer, a sample of the tumour was tested to identify how much of a particular protein called HER2 the cancer cells contained. Her Doctor discussed her case with the Multi-disciplinary Team at the hospital. (A Multi-disciplinary Team, or MDT, is a team of doctors and other health professionals with particular expertise in diagnosing and treating specific types of cancer. Histopathologists are key members of MDTs).

Sue's tumour was shown to be HER2-positive meaning that the cancer cells produced a lot of this protein. As well as surgery to remove the tumour, the MDT was able to advise that Sue was suitable for treatment with the drug Herceptin which is known to be particularly effective in treating HER2-positive tumours.

Points for discussion:

1. What are the advantages of having genetic information from people with a specific disease? Think about the use in scientific research.

2. What are the disadvantages of storing genetic information from a large number of people? Who might misuse the information?

The Royal College of Pathologists Pathology: the science behind the cure IBMS Institute of Biomedical Science

The Association for Clinical Biochemistry

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Ceri's story

Sue has been told that her breast cancer may be hereditary. This means that she may have passed down genes to her 22 year old daughter Ceri, which puts her at a higher risk of suffering from breast cancer in the future.

In order for Sue and Ceri to know for sure, Ceri would have to undergo a genetic screening test to look for these 'cancer causing' genes.

Points for discussion:

- 1. Do you think Sue should ask for Ceri to be screened?
- **2.** If you were in Ceri's place, would you want to know if you had inherited these 'cancer causing' genes?
- **3.** If scientists do identify these genes in Ceri, there are a few options available as she grows older:
- Avoid environmental factors which can increase your risk of cancer can you think of any?
- Increased 'surveillance' where Doctors would examine her on a regular basis with x-rays to detect any cancer early.
- 'Risk-reducing' surgery when she is older she could decide to have both breasts removed, usually followed by reconstruction surgery.

What do you think you would you do?