

CANCER GENETICS – FRIEND OR FOE OF THE JUNIOR DOCTOR?

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Recently there has been a surge in knowledge of cancer genetics, which has led to a rapid evolution of genetic counselling. Nowhere is this more so than in breast cancer where the importance of the BRCA1 and BRCA2 genes as predisposing factors has become widely recognised. Furthermore, the fact that tests for these genes are available has inspired a rapid increase in patient and relative demand for such services.

Some of the practical implications of this were dramatically brought home to me recently by a case on the Macmillan unit at Lancaster Royal Infirmary which demonstrated the particular dilemmas which can sometimes face the junior doctor. Our patient, a 66-year-old woman, had undergone a left mastectomy for pathologically proven breast cancer 22 years earlier and had recently re-presented with a malignant pericardial effusion and extensive mediastinal lymphadenopathy. Despite palliative chemotherapy she deteriorated rapidly. Her two sons, who were next of kin, were contacted as she was expected to die within a few hours.

It was at this point that genetic and ethical issues arose. Her sister had died from breast cancer which had presented at age 34 and her niece, who had suffered one previous benign breast lump and had been referred for genetic counselling, was concerned about future risk. We were requested, on the niece's behalf, to take a blood sample from the patient for genetic testing (BRCA1/2). Unfortunately at this point the patient was unconscious and could not give consent and died before this issue could be broached with her sons.

We thought that was the end of the matter but, after five days, late on a Friday afternoon when a long-planned walking weekend beckoned, I was contacted again by the family on behalf of the niece to ask if we had any stored samples from her aunt which would be suitable for genetic testing. Because

she was my patient and my consultant was unobtainable, I found myself coordinating the response.

It turned out that no suitable samples were available. I therefore contacted the Regional Genetics Unit in Manchester directly which suggested that liver or spleen samples from the deceased would be the most useful for extraction of DNA for the test. But there were problems. Was this ethical? Was it legal? On consultant advice, I sought out the trust lawyer. This was urgent as the deceased was due for cremation early on Monday morning. The advice given was that the samples should be obtained, but that we would need the written consent of the two sons.

The consultant pathologist agreed to take the samples in the hospital early on Monday morning. It turned out, however, that the body had already been released to the undertaker in Heysham. Further discussion suggested it was still best to take a sample. I counselled the sons to the best of my understanding and finally obtained their consent at 8.30am on the Saturday. My SHO colleague and I then attended the undertakers, suitably equipped to perform an open liver biopsy definitely not our intended Saturday morning activity! I ensured that the sample was frozen and later transported to the Regional Genetics Centre. All that remained was for me to contact the clinical geneticist who subsequently saw the family prior to testing. Finally my walking weekend could begin.

This was an extremely labour-intensive exercise, raising important economic, ethical and legal issues which, fortunately, could be properly addressed in this case. Such situations will become more frequent as public awareness increases and there may be a need for more widely available guidelines for junior staff.