Genetic Testing in Familial Breast Cancer

Information for referring clinicians

Your patient may have approached you with concerns about a family history of breast or ovarian cancer.
We have asked women who contact us directly to seek a referral through their family doctor or specialist.

Who should be referred?

- ☑ Women with breast cancer and a family history of breast or ovarian cancer.
- ☑ Women with a family history of breast or ovarian cancer, who are concerned about their risk.

What will happen after referral?

Upon receipt of your referral, the nurse co-ordinator will send your patient a questionnaire asking for details of their personal and family history of breast, ovarian and other cancers. This questionnaire is returned by post, and evaluated by the clinical team. Women judged to be at low risk will be reassured by letter, and will only be seen if they have continuing grave concerns. Women judged to be at high risk will be invited to attend one of the breast cancer clinics for further investigation and possible predictive testing.
The referring clinician will be kept informed of all relevant findings in the normal manner.

What kinds of genetic testing are possible?

Diagnostic genetic testing is used to identify mutations (changes in the DNA) in two genes called BRCA1 and BRCA2 which are present in some women with breast or ovarian cancer and a family history of breast or ovarian cancer. Finding such mutations has implications for the patient's own risk of further cancers. Finding a mutation also makes it possible to offer tests to adult relatives to see if they are at risk of developing cancer. This testing of healthy relatives is called *predictive genetic testing*.
NB: Currently (August 2004), The NCMG laboratories are not in a position to process new diagnostic testing requests pending the appointment of additional laboratory staff. New families are still being seen in the clinics.

Is predictive testing available to all high-risk women?

No. We can only offer predictive testing in families where we can identify a mutation in the BRCA1 or BRCA2 gene in an affected individual. This requires that a relative who has breast or ovarian cancer be available and willing to be tested, and that we can identify a mutation in one or other gene.

To whom should I refer my patient?

Women with breast or ovarian cancer and and/or a family history of either cancer should be referred to Professor Andrew Green at the National Centre for Medical Genetics.

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