

# Cancer Genetics Gazette

This gazette is sent to you from the Familial Cancer Unit  
of the South Australian Familial Cancer Service

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Welcome to the third issue of the Cancer Genetics Gazette for 2004. This newsletter aims to provide specialist clinicians with up-to-date information about familial cancer.

Please feel free to send suggestions for future topics as well as feedback about any articles that feature in the Cancer Genetics Gazette to:

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This newsletter features an article about **Screening for Breast Cancer for Women at High Risk** by

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## SCREENING FOR BREAST CANCER IN WOMEN AT HIGH RISK.

Women at potentially high-risk for developing breast cancer make up approximately 1% of the female population and are a group of mixed composition encompassing those with a proven mutation at the BRCA1 or BRCA2 gene sites and those with a specifically-defined personal or family history.

The lifetime risk of developing breast cancer is greatest in those women with genetic mutations and this risk reaches 80-90% (1). Cancers developing in these women occur at an earlier age than sporadic cases and by age 50, 50% of the BRCA1 and BRCA2 mutation carriers have developed the disease (1). Screening protocols must therefore begin at an earlier age than for low-risk women. This has been suggested at as early as age 30 (1) but NHMRC guidelines currently recommend 40 as the age to commence screening. In an individual case, screening should begin 5 years earlier than the age at diagnosis of the youngest affected first degree relative.

Younger premenopausal women have denser breast tissue than those aged 50 and over (2) and the sensitivity of mammography is reduced by this (3). Conventional screening protocols need to be altered in this group. Mammography remains the main screening tool as this is the only modality with a proven ability to reduce the death rate but additional procedures are often required.

Mammography has a sensitivity of 98% in detection of breast cancer in women of all ages with fatty breasts but this falls to 48% in the subgroup of women with the densest fibroglandular tissue pattern (2). Both young age (less than 50 years) and being in a high-risk group for developing breast cancer are independently associated with dense breast tissue patterns on mammography (2,4).

The addition of supplementary screening tests such as clinical examination, whole breast screening ultrasound (US), or magnetic resonance imaging (MRI), does not detect more cancers in women with fatty breasts (2,6)

but there is a 15% increase in cancer detection by adding whole breast US to mammography in patients with mammographically dense breast tissue (2).

A pilot screening MRI study with small sample numbers has shown an increased cancer detection rate of 3.8% when MRI is used to screen high risk patients with normal mammography and US studies (5). A recent programme of prospective breast cancer screening of women with a familial or genetic predisposition with both annual MRI and mammography has shown MRI to be more sensitive in detecting tumours but the lower specificity led to additional examinations and biopsies (7).

Both screening US and MRI are highly operator and protocol dependent and both are most effective in experienced hands. Screening MRI faces additional difficulties such as contrast enhancement in normal hormonally-stimulated breast tissue and localisation problems for biopsy and treatment of lesions that are discovered on MRI but are occult on MG and US (3). At this time breast MRI is used only as a diagnostic tool in specific, difficult clinical cases and, as yet, it has no role as a screening tool.

### **RECOMMENDED SCREENING PROTOCOL IN HIGH RISK WOMEN.**

1. Begin annual mammographic screening at 40 years, or 5 years before the age at diagnosis of the youngest affected relative.
2. Additional screening physical examination every 6-12 months.
3. Add annual screening whole breast US if a dense tissue pattern is present on the mammogram.
4. At this stage, MRI does not have a role in screening asymptomatic high-risk women.

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### **Audit group meetings**

In South Australia the main method for fostering multi-disciplinary management of familial cancer has been quarterly meetings of clinical, genetic, and laboratory experts. At these Audit Group meetings we review the indications for genetic testing, the outcome of recent laboratory studies, and the appropriate management of those shown to carry an abnormal gene. There are separate Audit Group meetings for familial breast and ovarian cancer, for familial colorectal cancer (including HNPCC) and familial neuro- & ocular-oncology (RB, VHL, NF2).

This model has worked well and has helped make the South Australian service among the best in the country. The Audit Group meetings are open to all professionals with involvement in the relevant cancers. If you would like to attend or to receive notices of meetings and minutes, please let me know.

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