

CEL 6 (2009)

26 January 2009

Addresses

For action

Chief Executives, NHS Boards
Chief Operating Officers, NHS Boards
Regional Genetics Units
Chairs/Lead Clinicians,
Regional Cancer Advisory Groups

For information

Directors of Public Health
Medical Directors, NHS Boards
Directors of Nursing, NHS Boards
Regional Cancer Network Managers
Professional Bodies/Other
organisations – see Annex

Enquires to:

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Dear Colleague

**CANCER GENETIC SERVICES IN SCOTLAND –
Management of Women with a Family History of Breast
Cancer**

The annex to this letter replaces the [HDL \(2007\) 8](#) which was issued in February 2007 and sets out core principles for the implementation of cancer genetic services. The working group was asked to reconsider the advice provided to clarify further the management of women at risk of familial breast cancer. The ovarian and colorectal cancer sections in the original [HDL \(2001\)24](#) remain extant.

The National Institute for Health and Clinical Excellence (NICE) in England published a [Clinical Guideline](#) in May 2004 for the classification and care of women at risk of familial breast cancer. This, as well as recognition of the scientific progress made since development of the original guideline, prompted a review by Geneticists in Scotland to see whether there was sufficient new evidence to support recommendations for change to the existing Scottish Familial Breast Cancer Guideline

NHS Boards should disseminate this updated guideline to relevant health care professionals and others.

Regional Genetics Units should, as set out in the revised guideline

- stratify individuals referred for advice into low, medium, high or very high risk groups
- further identify and counsel individuals requiring clinical surveillance, gene testing or family screening for inherited cancers

NHS Boards are expected to ensure clear referral pathways are in place locally for onward referral where appropriate from the Regional Genetics Unit to the relevant clinical cancer services.

Further copies of the revised guideline for the *Management of Women with a Family History of Breast Cancer* can be accessed on the *Scotland's Health on the Web* website at <http://www.show.scot.nhs.uk/>.

Yours sincerely

DR HARRY BURNS
Chief Medical Officer

DEREK FEELEY
Director of Healthcare Policy and Strategy

CANCER GENETICS SERVICES IN SCOTLAND

Professional bodies/other organisations to whom this letter and Annex have been sent are:

- Members of the Scottish Cancer Taskforce
- Members of the Scottish Genetic Advisory Group
- Royal College of Surgeons, Edinburgh
- Royal College of Physicians, Edinburgh
- Royal College of Physicians and Surgeons, Glasgow
- Royal College of General Practitioners, Scottish Council
- Royal College of Obstetricians & Gynaecologists
- Royal College of Radiologists
- Royal College of Pathologists
- College of Radiographers
- Royal Pharmaceutical Society of Great Britain, Scottish Department
- Academy of Royal Colleges and Faculties in Scotland
- British Medical Association, Scottish Office
- Scottish Joint Consultants' Committee
- Royal College of Nursing Scottish Board
- Scottish General Practitioners Committee
- Scottish Health Council
- Scottish Cancer Coalition
- Scottish Medical and Scientific Advisory Committee

Management of Women with a Family History of Breast Cancer

Guideline Summary

**Table 1. Breast cancer
Risk Stratification and Counselling**

Low Risk	Moderate Risk	High Risk	Very High Risk / Gene Carrier
<ul style="list-style-type: none"> • Anyone not fulfilling moderate, high or very high risk criteria 	<ul style="list-style-type: none"> • One first degree relative with breast cancer diagnosed under the age of 40, or one first degree relative with male breast cancer diagnosed at any age; • Two first or one first and one second degree relative with breast cancer diagnosed under 60, or ovarian cancer at any age, on the same side of the family; • Three first or second degree relatives with breast or ovarian cancer on the same side of the family where one is a first degree relative of the proband or of the proband's father; <p>A case of bilateral breast cancer should be treated as the equivalent of 2 affected relatives.</p> <p>In this context a first degree female relative is mother, sister or daughter. A second degree female relative is grandmother, granddaughter, aunt or niece.</p>	<ul style="list-style-type: none"> • Families with four or more relatives affected with either breast cancer under age 60, or ovarian cancer at any age, in three generations; Families where one individual has had both breast and ovarian cancer; • Families where there is an estimated 20% likelihood of a BRCA1, BRCA2 or p53 mutation; • Individuals with a lifetime risk of developing breast cancer of 30% or more. <p>The individual being assessed should be a first degree relative of an affected family member or a second degree relative through an unaffected male. Affected individuals should be first degree relatives of each other or related through unaffected males.</p>	<ul style="list-style-type: none"> • Female carrier of a mutation in BRCA1, BRCA2 or p53; • At 50% risk of carrying a mutation in BRCA1 or p53; • Women in their thirties whose 10 year risk is greater than 8% as assessed at age 30, or in their forties and whose 10 year risk is greater than 20% as assessed at age 40, or 12% where there is a dense mammographic pattern.

Table 2. Surveillance Guidelines

Low Risk	Moderate Risk	High Risk	Gene carrier or Very High Risk
<ul style="list-style-type: none"> • Reassurance. • Provision of Information. • National Breast Cancer Screening Programme from age 50. 	<ul style="list-style-type: none"> • Surveillance should be offered from the age of 40 or 5 years younger than the earliest age of onset of cancer in the family. • Mammography should be two yearly below age 40, and annually age 40 to age 50. • Mammography should not usually be commenced before age 35, and all women in this group should be offered mammography by the age of 40. • Breast examination should be offered where possible, and may be appropriate before the age of 35 where there is a family history of early onset cancer. <p>The surveillance programme should be audited as part of the national cancer genetics audit and where possible patients should be recruited into national studies to investigate the efficacy of such surveillance.</p>	<ul style="list-style-type: none"> • Surveillance should start at age 35 or 5 years younger than the youngest age of onset in the family. • Mammography should be 2 yearly below age 40, annually age 40 to 50, and subsequently 18 monthly from 50 to 70. • Mammography should usually start at age 35, and should not be offered under the age of 30. • Breast examination should be offered, particularly to women who are considered too young for mammography, who come from families where there has been onset of cancer before age 35. <p>Genetic testing should be offered in these families, if a sample is available from an affected relative. Where mutation testing cannot be offered, the possibility that the woman may be at sufficiently high risk to be offered MRI (see next column and risk table) should be considered.</p>	<ul style="list-style-type: none"> • Surveillance should start from age 25, or individualised according to the family. • Annual breast examination from 25 or 5 years younger than earliest age of onset, whichever is younger. • Mammography should be 2 yearly from age 30 to 39, annually from age 40 to 50, and subsequently 18 monthly from 50 to 70. • Breast MRI should be offered in addition to mammography, and should be annually from age 30-49. ^a Breast MRI should only be offered under 30 in rare very high risk situations eg p53 gene carrier. • Prophylactic mastectomy or, where appropriate, salpingo-oophorectomy should be discussed with the patient. <p>Bilateral mastectomy should be considered if a woman who is a gene carrier, or is at very high risk, is found to have a breast cancer.</p>

BRCA1 and BRCA2 Mutation Testing Criteria

Testing for mutations in BRCA1 or BRCA2 should be offered where there is a 20% likelihood of finding a mutation in one of these genes. The method of determining this risk should be recorded to allow effective audit of criteria used. Currently an Evans score of 20 or greater, or a family history covered by the empirical data published by Myriad, are the most appropriate ways of estimating mutation detection rate. There will be some clinical situations, outside these scoring systems, where the decision to test will be guided by clinical judgement.

Risk assessment to determine whether women are ‘very high risk’

Where a genetic test is not possible, the recommendation for breast MRI is determined by the 10 year risk of breast cancer, as assessed by use of a validated risk assessment tool such as Tyrer-Cuzick.