

## Joint position statement

# The role of single nucleotide polymorphisms (SNP) testing for personalised breast cancer risk prediction

As at 11 November 2013



## Joint position

At present, we recommend that the use of a SNP-based breast cancer risk assessment test should only be undertaken after an in-depth discussion led by a clinical professional familiar with the implications of genetic risk assessment and genetic testing, including the potential insurance implications.

We support efforts to educate the public about why genetic tests are best performed in a clinical setting that:

- provides genetic counselling by a suitably qualified professional
- allows personalised counselling and interpretation of test results, and
- implements legal provisions regarding privacy and health information.

We also recommend that any test which may be the basis for medical decision-making be performed by an accredited medical laboratory.

## What is SNP testing?

- Current SNP analysis typically involves testing for one or more common genetic variations that are each associated with a slight increase in cancer risk. These variations are known as single nucleotide polymorphisms (SNPs, pronounced "snips").
- Over 60 SNPs have been identified which can slightly increase a woman's risk of breast cancer, and the list continues to grow (Sakoda et al).
- SNP testing does not usually test for rare genetic variations that carry a high risk of developing cancer. For example, current SNP testing does not include complete analysis of the high-risk breast cancer genes, BRCA1 and BRCA2, that are analysed in some women with familial breast cancer.
- The interpretation of breast cancer SNP testing varies depending on the clinical setting of the person tested. While SNPs tests may provide useful cancer risk information in the future, their clinical utility at present is uncertain in our view. We support continuing research and development of such tests.

## Rationale

Clinical professionals recognise the importance of patients identifying and managing risk factors for disease such as lifestyle and family history.

As new technologies emerge that have a potential to assist in identification of genetic risk of cancer without family history, it is important for clinicians and patients to have access to information to assist them to understand the potential benefits and limitations of such tests.

The appropriate interpretation of a test assessing the risk of cancer requires an understanding of:

- **the analytical validity** of the test i.e. whether the test reflects the genetic status of the person tested
- **the clinical validity** of the test i.e. whether the test carries clinically meaningful information for the person tested
- **the clinical utility** of the test i.e. whether the test provides information that is not already available to the person tested
- **the ethical, social, and legal implications** of the genetic test result for the person having the test (for example, insurance implications).

## The availability of SNP testing for breast cancer risk

- Breast cancer SNP testing integrated with a personal and family history risk factor assessment of breast cancer is commercially available in Australia at a cost to the woman having the test.
- Breast cancer SNP testing is occurring within research studies at no cost to a participant but the timeframe of return of results is variable depending on the trial.

## Points to consider around SNP testing for breast cancer risk

- At this stage, the addition of SNP testing to a conventional personal and family history risk factor assessment offers only a modest gain in information and clinical relevance beyond that which is already available from a woman's personal risk factors and family history, especially in premenopausal women (Mealiffe et al 2010; McCarthy et al 2013; Husing et al 2012; Harlid et al 2012; Park et al 2012; Heald et al 2012; Campa et al 2011)
- There is the potential for a woman to erroneously believe that she has had a test for mutations in high penetrance cancer predisposition genes such as BRCA1 and BRCA2.
- SNP test results and the fact that a test has occurred will need to be disclosed to an insurance company in a similar manner to any other type of germline genetic testing.  
[http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/copy\\_of\\_FS23A.pdf](http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/copy_of_FS23A.pdf)

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## **About COSA**

The Clinical Oncology Society of Australia (COSA) is Australia's peak multidisciplinary society for health professionals working in cancer research, treatment, rehabilitation and palliative care with over 1600 members. COSA is recognised as an activist organisation whose views are valued in all aspects of cancer care. We are allied with, and provide high-level clinical advice to Cancer Council Australia.

## **About HGSA**

The Human Genetics Society of Australasia (HGSA) was formed by clinicians and scientists in 1977 to provide a forum for the various disciplines collected under the title of Human Genetics, including clinical genetics, cytogenetics, genetic counselling, molecular genetics, biochemical genetics, genetics in education and cancer genetics. HGSA is recognised as the peak professional body providing education and training (accreditation) to human genetics professionals.

## **About RCPA**

The Royal College of Pathologists of Australasia (RCPA) is the leading organisation representing pathologists and senior scientists in Australasia. Its mission is to train and support pathologists and senior scientists and to improve the use of pathology testing to achieve better healthcare.

The College was first established in 1956 and has been responsible for the training and professional development of pathologists since that time and more recently senior scientists. College members come from across Australasia including Australia, New Zealand, Hong Kong, Singapore, Malaysia and Saudi Arabia. The RCPA is also responsible for the promotion of the science and practice of pathology.

## **About RACGP**

The Royal Australian College of General Practitioners is Australia's largest professional general practice organisation and represents urban and rural general practitioners. The College represents over 20,000 members working in or towards a career in general practice. The College's mission is to improve the health and wellbeing of all people in Australia by supporting GPs, general practice registrars and medical students. This is achieved through education, training and ongoing professional development, including the development of resources and guidelines.