

# Genetics and Genomics Education Australia

KRISTINE BARLOW-STEWART | A/PROFESSOR



THE UNIVERSITY OF  
SYDNEY

MASTER OF GENETIC COUNSELLING  
SYDNEY MEDICAL SCHOOL, NORTHERN



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# PRINCIPLES FOR THE TRANSLATION OF 'OMICS'– BASED TESTS FROM DISCOVERY TO HEALTH CARE

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April 2015

| WORKING TO BUILD A HEALTHY AUSTRALIA |

National -  
NHMRC

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## Genetics and genomics resources for clinicians and researchers

### Health Topics

Alcohol Guidelines

Complementary medicines

Electronic cigarettes

Genetics and Genomics 

Genetics and genomics resources for consumers

Genetics and genomics resources for clinicians and researchers

Health effects of water fluoridation

Indigenous health

Nutrition

Obesity and Overweight

Parenting and child wellbeing

Lead blood levels

Talking with your health professional

Water Quality and Health

Wind Farms and human health

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The following resources have been developed by NHMRC to provide information and advice to clinicians and researchers on genetics or genomics and human health:

### Clinical Utility of Personalised Medicine

*Clinical Utility of Personalised Medicine* provides an overview of the role of genomics in personalised medicine, and its potential to improve health care. The document seeks to support health professionals in understanding the applications, utility and limitations of personalised medicine in clinical care.

> [Clinical Utility of Personalised Medicine](#)

### Medical Genetic Testing: Information for Health Professionals

*Medical Genetic Testing: Information for Health Professionals* provides information for use by health professionals involved in genetic testing. It supports the health professional in assisting patients with genetic tests and interpreting results in the context of clinical decision making.

> [Medical Genetic Testing: Information for Health Professionals](#)

### Principles for the Translation of 'Omics'-Based Tests from Discovery to Health Care

The NHMRC *Principles for the Translation of 'Omics'-Based Tests from Discovery to Health Care* are designed to assist researchers and clinicians in translating omics-based discoveries into validated tests that are clinically useful.

> [Principles for the Translation of 'Omics'-Based Tests from Discovery to Health Care](#)

### Discussing Direct-to-Consumer Genetic DNA Testing with Patients: A Short Guide for Health Professionals

*Discussing Direct-to-Consumer Genetic DNA Testing with Patients: A Short Guide for Health Professionals* provides health professionals with information to assist when patients present with Direct-to-Consumer genetic DNA tests or when patients wish to discuss the option of purchasing such tests.

> [Discussing Direct-to-Consumer Genetic DNA Testing with Patients: A Short Guide for Health Professionals](#)

### DNA Genetic Testing in the Australian Context: A Statement from the NHMRC

*DNA Genetic Testing in the Australian Context: A Statement from the National Health and Medical Research Council* provides health professionals and policy-makers information on DNA genetic tests that are used in the diagnosis of medical conditions and about an individual's risk of developing health-related conditions.

> [DNA Genetic Testing in the Australian Context: A Statement from the National Health and Medical Research Council](#)

### Biobanks Information Paper


The NHMRC *Biobanks Information Paper* provides information relevant to the establishment, management and governance of biobanks in Australia. The paper identifies best practice in regard to standardisation of biobank policies, practices and procedures based upon national and international literature.

> [Biobanks Information Paper](#)

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The following resources have been developed by NHMRC to provide information and advice to consumers on genetics or genomics and human health:

### The Human Genome

The human genome consists of the complete set of human genetic material that is contained in a human cell. In most human cells, the genetic material is made up of long DNA strands that are packaged into 23 pairs of chromosomes. A genetic disease or condition is caused by one or more genetic changes to the DNA code. This resource provides information about the human genome and how diseases or conditions can be caused by genetic mutations.

[> The Human Genome](#)

### Sequencing Your Genome

Rapid advances in DNA sequencing technologies now allow an individual's whole genome to be sequenced. Although this is still relatively expensive, it is becoming affordable and more readily available. This resource provides information for consumers about what it means to sequence your genome and what personal value it may or may not provide you.

[> Sequencing Your Genome](#)

### Epigenetics and Health

Epigenetics has recently become a popular explanation for complex diseases of unknown origin. Epigenetics is the study of mechanisms that control how genes are switched 'on' and 'off' without any changes in the actual DNA of the cells. This resource provides information about epigenetics and health.

[> Epigenetics and Health](#)

### Use of Genetic Information in Sport

DNA testing can be used for a number of purposes not related to health. One area of interest is the potential for DNA testing and genetic profiles in sport applications such as talent identification, individualised training programs and avoidance of sport-related injury. This resource provides information about the use of genetic information in sport.

[> Use of Genetic Information in Sport](#)

### Personalised Medicine and Genetics

Many of us wonder why some people develop cancer while others do not and why a medication might work well for one person but be less effective or cause serious side effects in another. Although these differences are due to a number of important factors such as age, weight and lifestyle, our genetic make-up also plays a part. Using information about genetic predisposition to disease is a key part of personalised medicine. This resource provides information about the use of genetics to predict disease development and to influence decisions about lifestyle choices or to tailor treatment to an individual.

[> Personalised Medicine and Genetics](#)

### Genetic Discrimination

Discrimination against an individual can appear in many forms. As DNA testing increasingly identifies differences in the genetic make-up of individuals, it becomes possible that people will be discriminated against based on genetic information. This resource provides information to consumers about genetic discrimination and how it applies in law and in health or life insurance.

[> Genetic Discrimination](#)

### Medical Genetic Testing: Health Information for You and Your Family

*Medical Genetic Testing: Health Information for You and Your Family* outlines the key issues that should be considered in relation to genetic testing. This resource provides information to assist people to:

- Understand the basics of DNA, genes and chromosomes;
- Consider whether to undertake medical genetic testing;
- Understand the potential outcomes of the genetic testing process; and
- Find further information and support.

[> Medical Genetic Testing: Health Information for You and Your Family](#)

### Understanding Direct-to-Consumer Genetic DNA Testing – An Information Resource for Consumers

In recent years, there has been significant increase in the number of Direct-to-Consumer (DTC) genetic DNA tests that consumers can purchase over the internet, without the involvement of a doctor. This resource has been developed to help consumers to better understand this type of testing. The NHMRC encourages individuals interested in DTC genetic DNA testing to exercise caution and consult their doctor if they choose to proceed.

[> Understanding Direct-to-Consumer Genetic DNA Testing – An Information Resource for Consumers](#)

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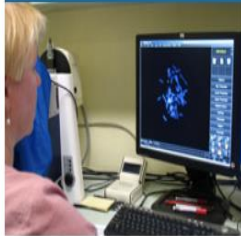


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## HGSA 40TH ANNUAL SCIENTIFIC MEETING



# Integrating Genomics into Healthcare

HGSA 40th Annual Scientific Meeting  
HOBART, TASMANIA • 6-9 August 2016

On behalf of the Local Organising Committee, we invite you to the Human Genetics Society of Australasia (HGSA) Annual Scientific Meeting to be held in Hobart, Tasmania, Australia on 6-9 August 2016.

The theme for the conference is "Integrating Genomics into Healthcare". We plan to explore the ways in which genetics and genomics are changing healthcare including the translation of genomics into diagnostics and targeted therapies as well as the psychosocial impact of genomic medicine on patients, families and communities.

The conference promises to be an exciting meeting with strong scientific content, covering various aspects of human genetics and genomics, including sessions around the following topics:

- Whole exome and genome sequencing for diagnostics and research
- Genetic therapies
- Bioinformatics
- Social and consumer impact
- Clinical phenotyping

hgsa.org.au



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eviQ home

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## Cancer Genetics

Welcome to the eviQ Cancer Genetics homepage.

### Family Cancer Clinics:

For information about family cancer clinics and services in your local area or other areas of Australia and New Zealand please see **Family Cancer Services, Centre for Genetics Education NSW Health** or the **Human Genetics Society of Australasia Find a Genetic Counsellor**

### New Referral Guidelines

**Referral Guidelines for Breast Cancer Risk Assessment and Consideration of Genetic Testing**

### New Risk Management Protocols

**Risk Management for Hereditary Papillary Renal Cell Carcinoma Type 1 (MET Gene)**

### Review Risk Management Protocols

- Risk Management for Li-Fraumeni Syndrome**
- Risk Management for Gorlin Syndrome/Nevoid Basal Cell Carcinoma Syndrome**
- Risk Management for Unaffected Male BRCA1 or BRCA 2 Mutation Carrier**
- Risk Management for Unaffected Individuals with a Heritable CDH1 Mutation**
- Risk Management for Lynch Syndrome**

### Review Genetic Testing Protocols

**Genetic Testing for Heritable Mutations in the STK11 Gene**

### Of interest :

The Cancer Genetics Reference Committee would like to advise they are aware of the following paper; however the group is awaiting further data analysis before making any amendments to eviQ protocols.

- Heemskerck-Gerritsen et al. Breast Cancer Risk After Salpingo-Oophorectomy in Healthy BRCA1/2 Mutation Carriers: Revisiting the Evidence for Risk Reduction JNCI 2015:107(5)

### eviQ Cancer Genetics GP Factsheet

**Recommendations for the management of breast cancer in women with an identified BRCA1 or BRCA2 gene mutation or at high risk of a gene mutation - Cancer Australia**

**BRCA1 and BRCA2 Variants of Uncertain Significance (VUS) - Further information for health professionals**

### Reference Committee Meetings and Email Notifications:



If you would like to participate in our reference committee meetings or subscribe to receive email notifications of new and updated protocols for Cancer Genetics, please **contact us**.


### Online professional development opportunities for rural health professionals

**Breast cancer in the family - what does it mean?** Is a NEW online module by Cancer Australia, available in October 2012. Developed in collaboration with the Australian College of Rural and Remote Medicine, it addresses the importance of assessing a family history of breast and ovarian cancer in your patients, and identifies risk-reducing strategies for women who are found to be at increased risk.

Utilising innovative online technology, Cancer Australia is increasing opportunities for general practitioners, nurses and other health professionals working in rural Australia to access information about the latest advances in **evidence-based breast cancer treatment and care** without the need to travel.

Registration is free- **access education now**

### Sub categories

-  [Referral Guidelines](#)
-  [Risk Management](#)
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### Acknowledgements

Select here to view the clinicians who have contributed to eviQ

### Quick Links

- [Cancer Australia](#)
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# NEW Information and Privacy Commission Guideline

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## Fact Sheets

Learn about genes, DNA, genetic patterns of inheritance and genetic conditions



## Family health history

Find out about the importance of knowing your family health history



## Pregnancy

Find out about health pregnancy preparation and testing options during pregnancy



## Cancer in the family

Information for those concerned about cancer in their family



### Genetic Services

- Genetic Counselling Services
- Genetic Testing Services
- Speciality Services
- Laboratory Services

### Links

- NSW Government
- NSW Health

### Sharing



# State-based - NSW

## genetics.edu.au



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## Individuals and Families

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[Newborn Screening](#)

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## Information for Individuals and Families

Here you will find everything from how to record your own family health history to learning basic facts about genes and DNA. Take a look through the following links for more ...

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[Understanding Genetics](#)

[What is Genetic Counselling](#)

[Genetic Conditions](#)

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# Health Centre for Genetics Education



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**Resources which are published and printed by the Centre can be ordered online**





## Health Professionals

Useful Resources

Referring to Genetics Services

Genetics in General Practice

Cancer

Pregnancy and Prenatal Testing

Policies and Clinical Guidelines

Learning Opportunities for Health Professionals

Newborn Screening

Pharmacogenomics

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## Health Professionals

Information and education for GPs and other health professionals

### IN THE SPOTLIGHT

**MTHFR genetic testing ~ Find out the facts**

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[Policies and Clinical Guidelines](#)

[Learning Opportunities for Health Professionals](#)

[Newborn Screening](#)

[Pharmacogenomics](#)



- › **Identifying gaps in genomic education:**
  - › **Genomic education programs**
    - Target group 1: Clinical Genetics Services
  - › *Genomics for Genetic Counsellors workshop Oct 2015*
    - Adapted from Training in Master of Genetic Counselling program
  - › *Clinical Genomic Data Analysis: (Garvan) Dec 2015*
    - Target group 2: Medical specialists- still planning
    - Target group 3: Nurses ++ (germline v tumour testing)
-

- › **Establish Genomic education section on Centre's website and GP tab:**
    - Point of care resources,
    - Programs for health professionals and patient information
  
  - › **Partnership with Genetic Alliance Australia:**
    - Rare Disease support groups
    - BRCA Information days
-



### Genetic Conditions

An extensive list of A-Z list of genetic conditions and information.

[Read more...](#)



### Support Groups

More than 50 different support groups connecting you with others.

[Read more...](#)



### GA's Projects

Take a look at our latest projects supporting genetic conditions.

[Read more...](#)

## ABOUT US

Genetic Alliance Australia, formerly Association of Genetic Support of Australasia (AGSA), a tax-deductible registered charity, was formed in 1988 to provide peer support and information for individuals and families affected by a rare genetic condition/rare disease.

- ✓ Contact other people affected by a genetic condition
- ✓ Personal stories
- ✓ How GA Australia can help you

## LATEST NEWS

### EVENT | BRCA Info Day UPDATE

18th October 2015

[... Details](#)

### NEWS | Breast cancer gene BRCA-1 cannot be patented, High Court rules

7th October 2015

ABC NEWS - The High Court has ruled the breast cancer gene BRCA-1 cannot be patented.... [Details](#)

## UPCOMING EVENTS

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### Genetic Conditions

An extensive list of A-Z list of genetic conditions and information.  
[Read more...](#)



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