

# ISO 15189 ACCREDITATION IN GENETIC TESTING LABORATORIES – AN AUSTRALIAN VIEWPOINT

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## Presentation Outline

NATA – who are we? What do we do?

Genetic testing in Australia – a profile

Genetic testing and accreditation – what are the challenges?

Has accreditation of genetic testing benefited the Australian public?

## NATA – AN OVERVIEW

- Established in 1947 by the Australian Commonwealth Government
- An association of members (laboratories)
- Sole national laboratory accreditation body recognised by MOU with Commonwealth Government
- Uses expertise of “peers” for technical assessment and advisory committees
- NATA staff are present at all assessments as Lead Assessors
- Currently, ~ 3,000 accredited laboratories across different fields of testing

## NATA – MEDICAL TESTING

Joint program with the Royal College of Pathologists of Australasia (RCPA)

Established in 1982 - voluntary program

Mandatory from 1986 for the payment of benefits from Medicare Australia  
(Federal government funded healthcare)

Accreditation covers all disciplines of pathology

Strong support from the National Pathology Accreditation Advisory Council (NPAAC)

Strong support from professional scientific bodies, e.g. Human Genetics Society of Australasia (HGSA)

Use of technical assessors and a Lead Assessor

## NATA – MEDICAL TESTING

Eighteen technical, four administration staff in Sydney, Melbourne and Brisbane offices

~ 600 accredited laboratories including eight overseas

Using ISO 15189 since 1 July 2005

Key stakeholders include the profession (through RCPA), Government, Scientific bodies, accredited laboratories and patients

Key role in the maintenance and improvement of safety and quality in pathology testing across Australia

Keen to maintain pace with the changing face of pathology to ensure accreditation continues to deliver a relevant, high quality, cost effective service

# GENETIC TESTING IN AUSTRALIA

Comprehensive study conducted by Dr Graham Suthers

*Familial Cancer Unit, Department of Genetic Medicine, Women's and Children's Hospital, Adelaide SA*

Released March 2009

**Laboratories were asked to provide information about tests which fulfilled all of the following criteria:**

- DNA- or RNA-based testing of human genes for medical purposes,
- Testing for heritable or non-heritable (somatic) genetic variants.
- The samples being tested were collected within Australia.
- The samples were tested during the 2006 calendar year.
- The testing was either performed in an Australian laboratory, or sent from an Australian laboratory to an overseas laboratory
- Testing was performed using non-Medicare funds.

**The Survey excluded tests which fulfilled any of the following criteria:**

- Testing done using Medicare funds.
- Medical testing of non-human genes (e.g. microbial genetic testing).
- Non-medical testing of human genes (e.g. paternity testing).
- Testing done principally for research purposes in relation to a specific project.
- Testing performed on samples received from overseas (including New Zealand).

## GENETIC TESTING IN AUSTRALIA

Number of laboratories – replies to the study

56 laboratories in Australia identified as performing non-rebateable molecular testing, 93% responded to the survey,

Range of testing

The study excluded tests which attracted a Medicare rebate e.g. FMR1 gene, Factor V Leiden, leukaemia markers, haemochromatosis (C282Y), HLA-B27)

> 430 different types of non-rebateable molecular tests offered by Australian laboratories

Types of laboratories

molecular genetics, genetic testing performed within mainstream laboratories e.g. haematology, (laboratories also performing testing in cytogenetics, biochemical genetics)

55% of tests identified in the study were offered by a sole laboratory within Australia

## GENETIC TESTING IN AUSTRALIA

Specialised reference laboratories

where are they? in all States

what tests do they offer? e.g Familial hemiplegic migraine (QLD), Frasier syndrome (VIC), Ashkenazi genetic disorders (VIC), late-infantile neuronal lipofuscinosis (SA), Marfan Syndrome (NSW), mitochondrial disorders (VIC)

The Human Genetics Society of Australasia (HGSA)

what role/s does a professional society play?

forum for collecting information – list of tests performed in different laboratories

provide advice to the accreditation body on matters raised at assessment

provide educational opportunities for members

working on registration criteria for medical scientists

## GENETIC TESTING AND ACCREDITATION

Can ISO 15189 apply to laboratories performing genetic testing?

Use of technical assessors

Proficiency testing for rare tests – how is this handled?

What are the issues we see at assessments?

## GENETIC TESTING AND ACCREDITATION

ISO 15189 – is a Standard for specific application in medical laboratories

- Management requirements
  - quality system, internal audits, control of documents, continual improvement mechanisms
- Technical requirements
  - staff training, pre-analytical processes, equipment, test methods, quality control, proficiency testing, reporting the test results

Activities within a diagnostic genetic laboratory can be accommodated within the framework of ISO 15189

Specific aspects of genetic testing not covered within the scope of the Standard

*Requirements for the Development and Use of In-House in Vitro Diagnostic Devices*  
NPAAC (2007 Edition) - based on Australian and International Standards

# GENETIC TESTING AND ACCREDITATION

## Technical assessors

- how are they appointed?
  - Identified by the Lead Assessor and/or Technical Assessor at assessment
  - Volunteer
  - Nominated by professional society
- the role of the pathologist – few molecular genetics pathologists
  - governance, clinical/laboratory interface, familiar with RCPA position on professional matters, Guidelines for reporting molecular genetic tests to medical practitioners RCPA Guideline *March 2009*
- the role of the scientist – expertise in testing methods, quality control, quality assurance, equipment etc

# GENETIC TESTING AND ACCREDITATION

Proficiency Testing (External quality assurance) – ISO 15189 Clause 5.6

Established programs

National: RCPAQAP, HGSA QAP

Participation – mandatory, together with the review and taking of corrective action

What happens when there IS no local QAP? Standard requires another mechanism, e.g interlaboratory exchange, overseas QAP

# GENETIC TESTING IN AUSTRALIA - ACCREDITATION

Class of test	Examples of tests	No of Accredited laboratories
Molecular Haematology	Factor V Leiden, thalassaemia -genetic abnormalities in $\alpha$ - or $\beta$ -globin chain synthesis; prenatal diagnosis sickle cell disease (Hb S) - detection of mutations in the $\beta$ globin gene; prenatal diagnosis	28
Molecular Immunology	HLA-B27, immunoglobulin Heavy Chain gene studies	10
Biochemical Genetics	Metabolite analysis, Enzymology, Newborn Screening	8
Cytogenetics	Blood, Bone marrow, Amniotic fluid, Chorionic villus tissue, Other tissues – non malignant, Other tissues – malignant, Fluorescent <i>in-situ</i> Hybridisation, Molecular karyotyping by microarray analysis	28
Molecular Genetics	DNA sequencing , Prenatal genetic testing, Pre-implantation genetic testing Genetic testing for constitutional gene variants (diagnostic and carrier testing) Predictive genetic testing Pharmacogenetic testing (results influence drug prescribing decisions) Genetic testing for mosaic gene variants (cancer and somatic mosaicism) Screening for an unknown mutation Assay for a defined mutation or polymorphism Assaying heterozygous loci Calculated estimate of risk of inheritance of an unknown mutation (Bayesian and linkage calculations)	39

## ACCREDITATION – THE BENEFITS

What has been the Australian experience?

recognition of the increasing development and discovery within the molecular world

acknowledgement that the accreditation process can deliver an increased confidence in the development and performance of testing

## ACCREDITATION – THE BENEFITS

### Benefits of accreditation

Use of international standards such as ISO 15189 provides a quality framework within which a laboratory can work and which can be used by an accreditation body to assess the service provided

Accreditation provides a way of determining, recognising and promoting the competence of facilities to perform specific types of tests

Facilities benefit from this because it allows them to find out whether they are performing their work correctly and to a benchmark level of quality and expertise

The assessment of facilities is carried out in a helpful, constructive manner that assists them in complying with the requirements for accreditation.