Strategic Plan 2010-2015

OFFICE OF POPULATION HEALTH GENOMICS

June 2010

EXECUTIVE SUMMARY

Population health genomics is a relatively new multidisciplinary field in which research findings in genetic and molecular technologies are integrated with evidence-based population health methodologies in order to prevent disease and improve individual and population health.

Population health genomics is founded on the principle that the human genome is a key determinant of health and susceptibility to disease. It follows that human disease occurs when environmental and lifestyle factors adversely impact on an individual's genome. Since genes are inherited through family and shared within communities, the science of population health genomics incorporates the deliberation of ethical, legal and societal issues.

This concept is reflected in the current international definition of population health genomics as 'the responsible and effective translation of genome-based knowledge and technologies for the benefit of population health[1].

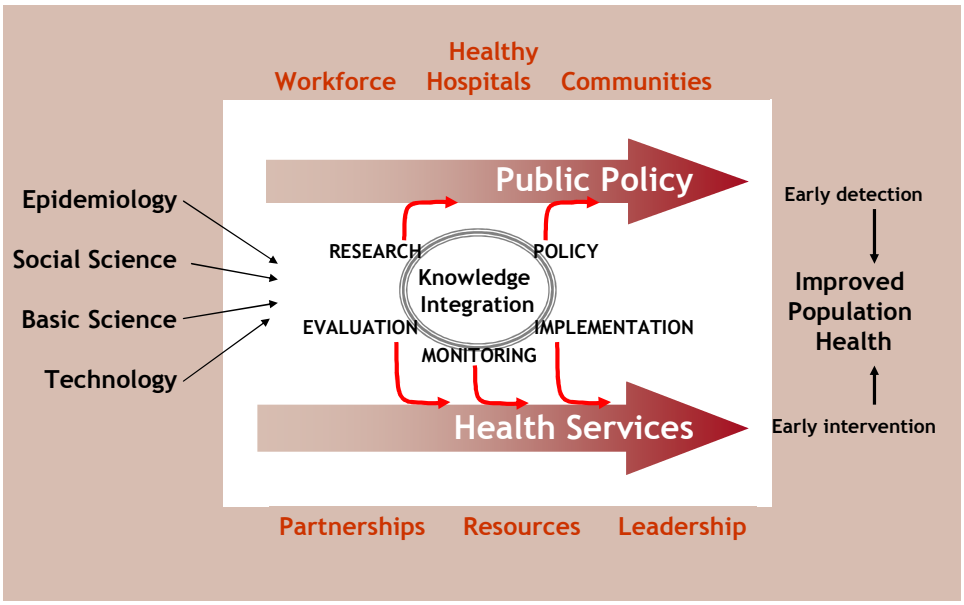
The primary goal of the Office of Population Health Genomics (OPHG) is therefore to integrate advances in genomics into health policy and improved health.

This strategic plan was developed with stakeholder consultation, community and expert opinion and outlines an operational framework for the planning, delivery and evaluation of genomic services in Western Australia between 2010 and 2015.

The elements of the strategic plan are to:

- Promote the development of an accessible, effective and co-ordinated genetic health service in Western Australia;
- Develop policy relating to genetic testing, screening programs and genetic technologies;
- Evaluate the clinical validity and utility of genetic service programs and assess performance of genetic services in Western Australia to determine the population health impact on genetic conditions;
- Develop an evidence-based population health policy framework for the implementation and governance of genetic services;
- Monitor and report on technological advances that might impact health provision, with particular emphasis on genetic and convergent genetic technologies including areas of cell and tissue engineering;
- Raise awareness of genetics in the community and healthcare sector to promote acceptance and integration of genetics into routine care;
- Lead and collaborate with peak leadership groups and community stakeholders.

[1] Burke W, Khoury MJ, Stewart A, Zimmern RL. Belagio Group: the path from genome-based research to population health: development of an international public health genomics network. Genet Med 2006;8:451-458.


Healthy
Workforce
Hospitals
Communities
Public Policy
Epidemiology
Social Science
Basic Science
Technology
RESEARCH
POLICY
Knowledge
Integration
EVALUATION
IMPLEMENTATION
MONITORING
Health Services
Early detection
Improved
Population
Health
Early intervention
Partnerships
Resources
Leadership

INTRODUCTION

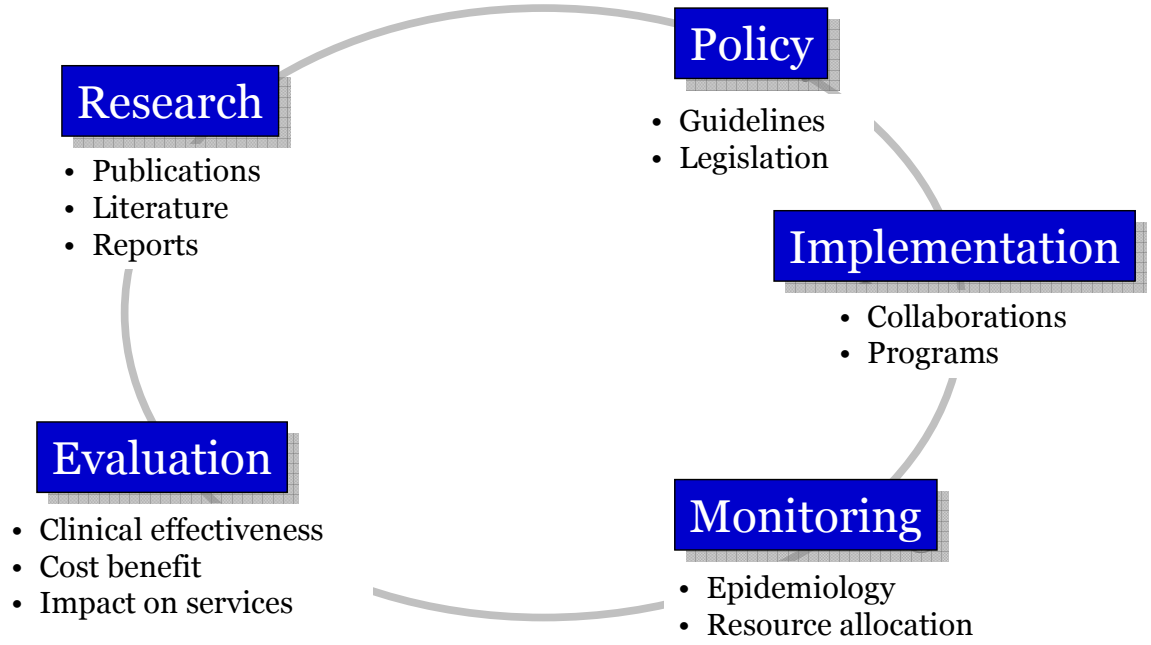
Genes control the structure and function of all cells within the body, ultimately influencing the health of an individual. Genetics is the study of inheritance, or the way that specific traits are passed from one generation to the next. A more encompassing field of study is genomics, which considers both the genetic constitution and environmental and lifestyle factors which can influence susceptibility to disease.

Once considered to be limited to rare paediatric disorders, genomics now finds application in many areas of modern medicine. For example, single gene and chromosomal disorders in Western Australia account for 2.6% of all admissions and 4.3% of total hospital costs for children and adolescents (Dye et al, 2010[2]).

The role of the Office of Population Health Genomics (OPHG) is to integrate advances in genomics into health policy and improved health. This is achieved by monitoring the cost-effectiveness of clinical genetic services and population genetic screening programs. OPHG has a mandate in Western Australia to evaluate clinical utility of new advances in molecular technology testing for disease prevention and health. In accord with these empirical approaches, OPHG supports deliberation of the ethical, legal and social issues arising from gnomic research by engaging with stakeholders in professional and community settings. Within clinical settings, OPHG provides policy advice on the development of genetic screening programs with a strong focus on population screening, targeted disease prevention initiatives and promotion of early detection to reduce the negative impact of disease.

[2] Dye DE, Brameld KJ, Maxwell S, Goldblatt J, Bower C, Leonard H, Bourke J, Glasson EJ, O'Leary P. The impact of single gene and chromosomal disorders on hospital admissions of children and adolescents: a population based study. Public Health Genomics 2010 (In press).

Figure 1: Policy Development Cycle



For more information about this strategic plan or about the Office of Population Health Genomics, please contact (08) 9222-6888 or genomics@health.wa.gov.au

Goals

1. Promote a cohesive and considered approach to advances in genomic technology;
2. Promote the development of an accessible, effective and co-ordinated genetic health services in Western Australia;
3. Assess the utilization and performance of genetic services and determine the population health impact of heritable conditions;
4. Identify best practices and develop an evidence-based policy framework for the implementation and governance of genetic services;
5. Raise awareness of genetics in both the community and healthcare sector to promote acceptance and integration into routine care.

DIRECTORATE OVERVIEW

Purpose

The aim is to integrate advances in genomics into health policy and improve health in the community.

Directorate History

The Office of Population Health Genomics (OPHG) was established as the Genomics Directorate in 2001 following a recommendation of the Health Administrative Review Committee Report (2001). The mandate was to develop the policy framework, strategic planning, resource allocation, program evaluation and quality assurance of genetic services within the health system.

OPHG has been influential in driving genetic policy development in Western Australia over the past ten years. Significant achievements have included:

- *Leading the Health Ministers" Advisory Council inter-jurisdictional committee inquiry into the impact of exclusive gene patents on diagnostic health services in Australia and recommendations for resolving that issue (2001-2008);*
- *Support the integrity of the statewide newborn screening program aims and activities, with special reference to storage and retention of blood samples (2000-2010);*
- *Establishment of the Western Australian Genetics Council (WAGC) (2002);*
- *Co-ordination of a Health Department response to the Australian Law Reform Commission/Australian Health Ethics Committee Inquiry into Protection of Human Genetic Information (2002);*
- *Appointment of the Chair in Medical Genetics (2003);*
- *Expansion of the Newborn Screening Program (2004) through the funding of a tandem mass spectrometer;*
- *Hosting the GenETHICS essay competition for high-school students in Western Australia (2004-2007);*
- *Evaluation of prenatal screening for Down Syndrome, genetic testing for familial cancer and genetic counselling services (2005);*
- *Participation in the national policy evaluation of evidence to support the introduction of mandatory fortification of bread-making flour with folate (2004-2008);*
- *Co-ordination of a pilot program to identify patients and affected relatives with familial hypercholesterolaemia (2007-10) supported by the Australian Better Health Initiative;*
- *Evaluation of prenatal screening practices (2005 -2010);;*

- *Design and execution of a deliberative democracy process to engage community and stakeholders in the development of Biobanks policy in Western Australia (2008-2009);*
- *Legislative change to introduce mandatory reporting to the Western Australian Birth Defects Register (2009-2010);*
- *Hosting the inaugural GRaPHInt International Conference on population health genomics (2009;.*
- *Endorsement and launch of guidelines for the establishment, management and operation of biobanks in Western Australia (2010).*

In the interests of knowledge dissemination, the Directorate is also committed to the publication of research findings in peer-reviewed, scientific journals. Thirty nine research papers have been published up to May 2010 (Appendix One).

Structure of the Directorate

Outcomes are devolved through two operational branches who share responsibility for these tasks:

- *Service Planning Branch* – which evaluates advances in genetic technologies for implementation into clinical practice;
- *Service Evaluation and Monitoring Branch*– which monitors genetic service delivery and engages the community to develop evidence-based policies.

Director	Dr Peter O'Leary PhD, MAACB, AFACHSE, ARCPA
Administrative Assistant	Jackie Ridler
Service Evaluation & Monitoring Branch	Caron Molster (Manager) BBis (Hons)
	Suzy Maxwell BA (Psych & Anthrop) GDipMarketing, MPH (Candidate)
	Sarah Baxendale BHealthSc, MPH
Service Planning Branch	Dr Hugh Dawkins (Manager) PhD
	Leanne Youngs BApplSc
	Joanna Brisbane BSc, MPH (candidate)

GOALS

1. PROMOTE A COHESIVE AND CONSIDERED APPROACH TO ADVANCES IN GENOMIC TECHNOLOGY

The advances in genomics research provide opportunities for improvements in clinical and population health. The development of molecular technologies has enabled faster, better and less expensive diagnostic pathways and investigations of genetic variants associated with diseases. The practice of personalised medicine is evolving rapidly raising the prospect of individualised prevention and treatment interventions that produce population health benefits. As scientific discoveries open greater opportunities for interventions, there is a need to engage with the social sciences to translate genomic information into a framework that addresses behavioural, lifestyle, legal and ethical principles.

EVIDENCE OF NEED[3]:

- 2.8% of all admissions to WA hospitals for children aged 0-19 are due to single gene and chromosomal conditions;
- Patients with single gene and chromosomal disorders have a higher number of admissions per patient and a longer mean length of stay compared to other patients;
- The cost per patient and cost per admission is higher for patients with single gene and chromosomal disorders than for other patients, accounting for 5.1% of the cost of hospital admissions of 0 – 19 year olds in WA (2006).

OBJECTIVES

1.1. Promote the integration of population health genomics in relevant local, State and National agencies

- Facilitate activities to promote the integration of genomics into clinical care through collaboration with partner agencies, organizations and programs;
- Support a State genetics advisory committee (the Western Australian Genetics Council (WAGC)) and relevant subcommittees;
- Establish and maintain partnerships across networks of clinical, laboratory, community and academic fields at both State and National levels.

1.2. Ensure prompt and appropriate State responses to National recommendations for issues relevant to genetic service delivery

- Monitor new recommendations for best-practice in the field of population health genomics;
- Ensure alignment of genomics policies with State Government priorities and policies;
- Monitor health outcomes following National initiatives such as folate fortification of staple foods, privacy laws and best-practice guidelines;
- Participate in the development of National policies for genetics in healthcare, including those for rare disorders.

[3] Dye DE, Brameld KJ, Maxwell S, Goldblatt J, Bower C, Leonard H, Bourke J, Glasson EJ, O'Leary P. The contribution of single gene and chromosomal disorders to pediatric hospitalizations in Western Australia. (In press) Public Health Genomics. 2010

1.3. Evaluate new genetic technologies for integration into clinical practice

- Monitor advances in genomics and their application to population health;
- Develop methods to address the population health implications of new genetic tests and technologies;
- Consider the ethical, legal and social implications of current and emerging genetic technologies.

2. PROMOTE THE DEVELOPMENT OF AN ACCESSIBLE, EFFECTIVE AND CO-ORDINATED GENETIC HEALTH SERVICES IN WESTERN AUSTRALIA

Genetic service delivery should allow equitable access to genetic and diagnostic testing with due consideration given to planning for future needs. This will occur through the integration of several health services including clinical genetics, genetics policy, genetic counselling and laboratory services. The development of and adherence to performance measures forms a critical aspect of implementation.

EVIDENCE OF NEED:

- Genetic disorders and congenital abnormalities occur in
  - 2%-5% live births (600-1500 babies in WA) (WABDR)[4],
  - account for 2.6% of admissions and 4.3% of total hospital costs[5]
- Determination of the burden of genetic disease is critical to inform delivery of newborn screening services and enabling evidence based capability and capacity building in genetic services.

OBJECTIVES

2.1. Promote availability of a comprehensive genetics service throughout Western Australia

- Identify outcome measures to demonstrate the effectiveness of genetic services;
- Determine actual costs per patient seen for different types of caseloads at both Genetic Services of Western Australia (GSWA) and outreach clinic sites;
- Promote continued viability of statewide clinical services by advocating for supplemental financial support as needed;
- Document the current infrastructure and capacity for delivering services and building capacity for future development.

2.2. Review and assess performance standards for the Newborn Screening Program

- Promote the program so that all infants receive screening in accordance with established guidelines and that screen-positive infants are identified, contacted and referred within an accepted timeframe;
- Develop a comprehensive program to provide education and in-service training about the newborn screening process to hospitals, physicians and midwives who deliver infants;
- Monitor the development of systems to link the newborn screening database with vital records on a continual basis in order to identify unscreened infants in a timely manner and to monitor long-term treatment and outcomes;
- Use existing population health data systems to determine utilization of newborn screening health services;
- Measure service performance against current best-practices.

2.3. Facilitate establishment of genetic registries to assist testing and clinical care for specific rare genetic diseases

- Explore the feasibility of extending the National pilot scheme to establish a registry and genetic laboratory services for Duchenne muscular dystrophy;

[4] Bower, C, Rudy, E, Callaghan, A, et al. Report of the Birth Defects Registry of Western Australia 1980 - 2008. http://www.kemh.health.wa.gov.au/brochures/health_professionals/wnhs0260.pdf. 2009.

[5] Dye DE, Brameld KJ, Maxwell S, Goldblatt J, Bower C, Leonard H, Bourke J, Glasson EJ, O'Leary P. The contribution of single gene and chromosomal disorders to pediatric hospitalizations in Western Australia. (In press) Public Health Genomics. 2010

- Co-ordinate antenatal and prenatal screening for haemoglobinopathies through the establishment of a registry for Western Australia and the development standards of care;
- Development of a rare disease strategy for WA and a Rare Disease Registry;
- Develop and manage a Prenatal Diagnostic Registry.

2.4. Reduce the population health burden related to preventable chronic diseases with a significant genetic component

- Promote the use of family history for genetic risk assessment of common chronic conditions.
- Utilise existing health databases to examine morbidity and mortality related to genetic conditions and assess the costs of medical care for these conditions.

2.5. Develop methods to assess the use of genetic services by the community

- Develop the tools to determine if there is equitable access to genetic services across the State and determine risks of not providing such services.
- Monitor whether referrals are appropriate for genetic consultations and implement strategies to address where necessary;
- Develop and implement methodologies for tracking the use of genetic screening and diagnostic services including counselling and test uptake;
- Determine adherence to best-practice guidelines relevant to genetic services;
- Monitor health outcomes of people with genetic disease.

2.6. Monitor standard of Prenatal Screening Program for Down Syndrome

- Assess utilization of prenatal screening program and ascertain case detection rate, test efficacy and further testing outcomes;
- Develop and implement a Model of Care for prenatal screening in Western Australia and monitor adherence to best-practices.

2.7. Monitor the incidence of neural tube defects as a result of mandatory fortification of bread-making flour with folic acid

- Measure base-line and ongoing folate consumption in both Indigenous and non-Indigenous populations through assessment of folate levels in the blood;
- Review data on the annual incidence of neural tube defects in Western Australia following mandatory fortification of bread-making flour with folic acid.

3. IDENTIFY BEST PRACTICES AND DEVELOP AN EVIDENCE-BASED POLICY FRAMEWORK FOR THE IMPLEMENTATION AND GOVERNANCE OF GENETIC SERVICES

An effective policy development process allows input from different perspectives, is flexible, includes extensive consultation and appropriate transparency, leads to informed decision making and will lead to the best outcome in the particular circumstances for those involved or affected.

EVIDENCE OF NEED:

- Evidence-based policy and guidelines are rare;[6]
- The key challenge in developing evidence-based genomics guidelines is to assess the clinical validity and utility of a application; and [7]
- Development of sound genetics guidelines requires timely and coordinated policy development process that builds on evidence based science and on-going community consultation.[8]

OBJECTIVES

3.1. Provide policy direction for the implementation of genetic services

- Develop policies, guidelines and position statements that are consistent with State government requirements, are evidence-based and demonstrate understanding and consideration of stakeholder and citizen viewpoints;
- Provide policy advice to stakeholders and government at a local, state and national level through participation on committees and working groups.

3.2. Optimise genetic testing services on the basis of clinical validity and utility

- Support an evidence base for introduction of new screening tests within the newborn screening program and for other proposed genetic tests;
- Evaluate the cost-effectiveness of proposed genetic tests.

3.3. Engage community and stakeholders to seek input into the policy, planning and design of genetic services

- Educate the community on matters of policy relevance;
- Develop tools and methodology to ascertain and incorporate stakeholder input into genetic policy development;
- Engage community and service providers to decrease the barriers inhibiting utilization of genetic services;
- Engender trust and engage community and stakeholder involvement in health service development;
- Provide reassurance to the community that genetic information will be appropriately managed and safeguarded.

3.4. Implement the WA State policy on retention and storage of newborn screening dried blood spot samples

- Review the current storage and retention policy for newborn screening blood samples;
- Promote the retention of samples for a defined period in a manner that preserves their integrity for DNA or other types of analysis;

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[6] Khoury et al. 2007. The continuum of translation research in genomic medicine: how can we accelerate the appropriate integration of human genome discoveries into health care and disease prevention? Genetics in Medicine, 9(10), p665-674

[7] Rogowski et al. Challenges of translating genetic tests into clinical and public health practice *Nature Reviews Genetics* 2009;10:489-495.

[8] Brand et al. The impact of genetics and genomics on public health. Eur J of Hum Genetics 2008;16: 5-13.

- Promote access to newborn screening specimens for research is approved through Health Research Ethics Committees;
- Assure privacy protections for data management and reporting newborn screening results to physicians, medical management centers and others.

4. RAISE AWARENESS OF GENETICS IN BOTH THE COMMUNITY AND HEALTHCARE SECTOR TO PROMOTE ACCEPTANCE AND INTEGRATION INTO ROUTINE CARE

A vast amount of genetic information is now available; however knowledge levels remain low in both the community and health profession. Improving population awareness and understanding of genetics are necessary prerequisites to the acceptance of genetic technologies by the community. An increased awareness of genetics in mainstream medical services will also improve the management of familial diseases and may foster innovative research projects and collaborations between medical professionals.

EVIDENCE OF NEED:

- Population health initiatives have demonstrated the gap between population health policy and community expectations.
- 28% of GPs regularly encounter patient demand for information about genetic disease in their practice particularly in regard to pregnancy or family planning[9];
- Despite this demand, GPs report a lack confidence in their ability to give advice to couples requesting genetic testing, and encounter difficulties explaining genetic information in lay terms and conveying risk concepts and the non-definitive nature of screening tests to patients[10];
    - < 19% of GPs expressed confidence in providing advice to a couple requiring information about prenatal thalassaemia testing, while 43% expressed low levels of confidence[11];
- A survey of GPs from New South Wales, found that 83% lacked confidence in offering genetic counselling[12];
- Most GPs admitted that their knowledge of genetics was extremely poor or inadequate and that they lacked confidence in counselling patients with regards to genetic issues[13].

OBJECTIVES

4.1. Prepare and maintain appropriate educational materials to support clinical genetics services

- Evaluate community and health professional knowledge levels relating to genetic disease and technologies;
- Collaborate with genetic service providers to determine education resource requirements and dissemination strategies;
- Monitor distribution and utilization of education resources;
- Review and evaluate existing education resources at periodic intervals;
- Increase the availability of culturally sensitive, educationally appropriate and scientifically accurate information about genetic conditions, risks and services;
- Provide advice to health professionals on genetic resource development as required;

[9] Metcalfe, S., et al., Needs assessment study of genetics education for general practitioners in Australia. Genetics in Medicine, 2002:4:71-77.
[10] Metcalfe, S., et al., Needs assessment study of genetics education for general practitioners in Australia. Genetics in Medicine, 2002:4:71-77.
[11] Qureshi NS. Armstrong, Modell B. GP's opinions of their role in prenatal genetic services: A cross-sectional survey. Family Practice, 2006;23:106-110.
[12] Huang QL, Trevena, McIntosh J. GP's experience and attitudes towards new genetics: Barriers and needs. Australian Family Physician, 2004;33:379-380.
[13] Huang QL, Trevena, McIntosh J. GP's experience and attitudes towards new genetics: Barriers and needs. Australian Family Physician, 2004;33:379-380.

- Maintain protocols for the development of health resources and ensure adherence with WA Health and State Government guidelines.

4.2. Promote awareness of OPHG activities and genetic services and resources amongst the community, health care providers and stakeholders

- Maintain a presence on the internet and establish links on relevant third-party websites;
- Distribute a brief review of Directorate activities via a newsletter to stakeholders;
- Participate in conferences and education events.

4.3. Ensure sustainability of genetic services by assisting in workforce development, education and co-ordination

- Liaise with genetic service providers and other relevant organisations to determine workforce requirements and training needs in genetics;
- Collaborate with providers of genetics education programs, including academic institutions, research organisations and education departments, to address training needs in genetics.

APPENDIX ONE

PUBLICATIONS OF THE OFFICE OF POPULATION HEALTH GENOMICS

Dye D, Youngs L, McNamara B, Goldblatt J, **O'Leary P**. The disclosure of information: a human research ethics perspective. Journal of Bioethical Enquiry 2010;7:103-109.

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O'Leary PC, Maxwell SJ, Youngs LM, Brameld KJ, Walpole IR. Outcomes of a cystic fibrosis carrier testing clinic for couples. Medical Journal of Australia (Letter to the Editor), 2010;192(4):238.

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Australian & New Zealand Journal of Obstetrics & Gynaecology. 2008; 48:492-500.

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Makepeace AE, Bremner AP, **O'Leary P**, Leedman PJ, Feddema P, Michelangeli V, Walsh JP. Significant inverse relationship between free T4 concentration and body mass index in euthyroid subjects: differences between smokers and non-smokers. Clinical Endocrinology 2008;69:648-652.

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