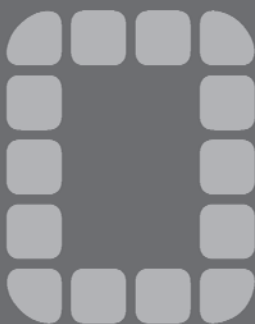


Office of Population Health Genomics

Annual Report 2008-2009



Director: Dr Peter O' Leary

Epidemiology & Health Economics

Genetic Burden of Disease program to monitor the impact of hereditary and genetic diseases on the services provided by WA Health.

Reference information on hospitalisations for single gene and chromosome disorders has been prepared. This dataset includes the number of people admitted to WA hospitals, number of admissions, length of stay and costs and percentage of familial admissions for 2000-2006.

This study will provide vital data for informed policy development and forward planning of health services and resources, as patients with genetic conditions represent an important portion of the population with special health care needs. Previous studies in other countries have found that patients with genetic conditions have more admissions, longer hospital stays and increased morbidity and mortality than patients without genetic conditions. In addition, improved care and therapeutic advances have resulted in increased longevity for many genetic conditions and, as a result, the health system will face new demands from these patients.

Mandatory folate fortification of food to reduce neural tube defects

This project involves co-ordination of studies to measure baseline blood folate and vitamin B12 levels and dietary folate levels in samples of the indigenous and non-indigenous population in WA prior to mandatory fortification of bread making flour with folate in October 2009. Data have been collected in the indigenous population in Broome and arrangements are underway to collect data in Balgo and at Derbarl Yerrigan clinics in Perth. Non-indigenous data collection is underway in the metropolitan area.

Expansion of newborn screening for congenital adrenal hyperplasia.

OPHG has continued to monitor the technical details of testing for CAH as part of the newborn screening program in conjunction with the newborn screening laboratory. We are also monitoring international developments with regards the cost-effectiveness of screening for CAH.

The storage and retention of newborn screening samples and personal information and potential use for secondary purposes (research, forensic DNA identification or in law enforcement).


In Australia, there is controversy about the appropriate length of time to store Guthrie cards, and whether they should be used for secondary purposes, such as research, DNA victim identification in natural disasters or in law enforcement. The secondary use of newborn screening cards raises several complex issues, with the fundamental concerns involving informed consent, privacy of information and the legal status of stored samples. The primary purpose of screening - the early diagnosis of treatable inherited disorders, needs to be balanced carefully against any requirement to obtain fully informed consent to retain newborn screening cards for other potential secondary uses.

Evaluation of population carrier testing for cystic fibrosis genes as preparation for a policy position.

An economic evaluation of cystic fibrosis prenatal carrier screening is being undertaken to establish the cost effectiveness of different models of screening. This will inform the development of WA Health policy on CF prenatal carrier screening.

Abstracts

Kate Brameld, Suzy Maxwell, Danielle Dye, Peter O'Leary, Jack Goldblatt, Carol Bower, Helen Leonard, Jenny Bourke, Emma Glasson. Measuring the impact of genetic disease in the WA population. [Abstract 175] Journal of Twin Research, May 2009.



Suzy Maxwell, Leanne Youngs, Kate Brameld, Elizabeth Geelhoed, Peter O'Leary. Economic evaluation of cystic fibrosis prenatal carrier screening. [Abstract 176] Journal of Twin Research, May 2009.


Kate Brameld, Peter O'Leary, Robert Cocciolone. An evaluation of prenatal screening strategies in the Australian context [Abstract 182] Journal of Twin Research, May 2009.

Papers published

O'Leary P, Lewis B. Maternal fetal screening. Chapter: in Methods, Kaplan L, Pesce A, Clinical Chemistry, 5th edition. Accepted for publication, 2009.

Brameld K, Dickinson J, Bower C, Goldblatt J, Murch A, Hewitt B, Stock R, O'Leary P. Association of first-trimester maternal serum PAPP-A, free-beta subunit human chorionic gonadotropin concentration, nuchal translucency and the combined risk measurement for Down's Syndrome with adverse pregnancy outcomes. Accepted for publication, Australian & New Zealand Journal of Obstetrics & Gynaecology 2008;48:529-535.

Cocciolone R, Brameld K, O'Leary P, Haan E, Muller P, Shand K. Combining first and second trimester markers for Down syndrome screening – think twice. Australian & New Zealand Journal of Obstetrics & Gynaecology 2008;48:492-500.



Prevention and Translation

Familial Hypercholesterolaemia Cascade Screening Pilot Program: a Flagship project of the Australian Better Health Initiative.

Familial hypercholesterolaemia (high blood cholesterol) cascade screening WA is a pilot program to identify index cases and to cascade screen relatives.

Over 130 index cases have been identified and over 200 relatives have been screened for FH, by early 2009.

The Familial Hypercholesterolaemia Clinic aims to screen 150 relatives by 30 June 2009. Patients identified with the conditions will be offered appropriate medical and dietary treatments to reduce their risk of cardiovascular disease and then returned to their GP for ongoing care.

OPHG is about to commence an evaluation phase of the program that will include a cost-effectiveness assessment, examination of clinical outcomes and genetic test data. A comparison of software data management options will also be addressed.

A Community Support Group has been established to provide mutual support and opportunities for families to learn more about this inherited condition.

A model of care for screening adults and children at risk of familial hypercholesterolaemia has been endorsed by the State Health Executive Forum and adopted by the Atherosclerosis Society of Australasia for nationwide dissemination. The program is currently being extended to include regional areas of the State.

Professional genetics education for allied health staff and GPs, including targeted community initiatives and collaborations with non-government organisations to increase awareness of family health history in chronic disease.

GP education initiatives were undertaken in partnership with the Osborne Division of General Practice and the Perth Primary Care Network as part of the West Australian Familial Hypercholesterolaemia program (FHWA). OPHG has achieved RACGP accreditation for education related to familial hypercholesterolaemia and Family Health History education modules.

Abstracts

Samantha Poke, Gerald Watts, Suzy Maxwell, Kate Brameld, Peter O'Leary. Familial Hypercholesterolaemia (FH) pilot cascade screening project. [Abstract 174], Journal of Twin Research, May 2009.

Suzy Maxwell, Caron Molster, Samantha Poke, Peter O'Leary. Community views on the communication of FH genetic information within families [Abstract 177]. Journal of Twin Research, May 2009.

Jennifer Hancy, Danielle Dye, Joanna Brisbane, Taryn Charles, Amanda Samanek, Peter O'Leary. Developing a genetics module for Year 10 Human Biology [Abstract 179]. Journal of Twin Research, May 2009.

Gaenor Kyne, Caron Molster, Sam Poke, Jennifer Hancy, Joanna Brisbane, Peter O'Leary. Your best chance of a healthy future may be knowing about the past: developing effective tools for the public to collect their family health history. [Abstract 187] Journal of Twin Research, May 2009.

Sarah Baxendale, Sally Appleton, Peter O'Leary. Evaluation of the prenatal screening and diagnostic tests patient information booklet. [Abstract 193] Journal of Twin Res. May 2009.

Papers published

Maxwell S, Molster C, Poke S, O'Leary P. Communicating genetic information within families. In press, Genetic Testing, 2009.

Reid GT. Walter FM. Brisbane JM. Emery JD. Family history questionnaires designed for clinical use: a systematic review. Public Health Genomics. 12(2):73-83, 2009.



Social and Community Research

Biobank community consultations on the protection and health benefits of deoxyribonucleic acid (DNA) sample collections held in WA Biobanks.

In August and November 2008, innovative deliberative community forums were held on “Biobanking in WA” with members of genetic support groups and interested citizens drawn from the WA community. Participants were provided with a range of perspectives on the issues then produced a set of recommendations for how biobanks should operate in WA. The recommendations have been considered and incorporated in the development of a governance and legal framework for biobanking in WA.

Abstracts

Leanne Youngs, Hugh Dawkins, Caron Molster, Peter O’Leary. Stakeholder engagement as an integral part of biobanks policy development [Abstract 180] *Journal of Twin Research*, May 2009.


Fiona Hope, Caron Molster, Peter O’Leary. Persistent disagreement about biobanks and the impact on public policy arising from public and consumer group engagement. [Abstract 182] *Journal of Twin Research*, May 2009.

Gaenor Kyne, Caron Molster, Peter O’Leary. Community engagement for policy development: are health consumers representative of the general public? [Abstract 186] *Journal of Twin Research*, May 2009.

Caron Molster, Bev McNamara, Taryn Phillips, Ayla Potts, Peter O’Leary. Evaluating deliberative public engagement from multiple perspectives: is it an effective methodology for genomic public policy development? [Abstract 199] *Journal of Twin Research*, May 2009.

Papers Published

Molster C, Charles T, Samanek A, O’Leary P. Australian study on community knowledge of human genetics and health. *Public Health Genomics* 2009;12:84-91.



Biotechnology, Ethical Legal

National Duchenne and Becker Muscular Dystrophy Register and gene sequencing.

OPHG has been appointed to lead the development of an options paper for the Clinical, Technical and Ethical Principle Committee of the Australian Health Ministers' Advisory Council on the establishment of a Muscular Dystrophy Register. Parent Project Australia, a voluntary organisation advocating for families affected by Duchenne (DMD) and Becker Muscular Dystrophies (BMD), is promoting a national register linked to full dystrophin gene sequence data. The impetus for a voluntarily register of all boys with this debilitating and ultimately fatal disease is to provide Australian families with the opportunity to participate in international clinical treatment trials arising from advances in genetic technologies and gene therapy.

Indigenous Chronic Disease Survey

OPHG has led the development of a strategy and implementation plan to help close the gap in life expectancy between the indigenous and non-indigenous populations, more widespread screening and follow up. The first stage has been to investigate and develop recommendations around IT systems to enable systematic and opportunistic screening, disease registers and recall processes.

National organ and tissue donation and transplantation

Though involvement with the Cognate Committee on Organ and Tissue Donation, OPHG contributed to the transition from "Australians Donate" to the new federally funded Australian Organ and Tissue Donation and Transplantation Authority. Specific activities included the responsibility we shared with NSW Health to deliver the Australian Paired Kidney Exchange Program (AKX) Guidelines.

Genes for Health Conference

The program for the joint conference of the Human Genetics Society of Australasia (HGSA) and the Genome-based Research and Population Health International network (GRaPH-Int) is almost finalised. The conference will bring together international experts in the field of basic genetic science, genetic testing, bioethicists, educators, genetic service providers, public health professionals, health service researchers, social marketers, primary care practitioners, policy makers and the public (<http://www.graphint.org/australia2009>).

Submissions

Submission to the Federal Senate Community Affairs Committee Inquiry into Gene Patents for the Government of Western Australia.

Papers published

Metcalfe SA, Bittles AH, O'Leary P, Emery J. Australia: Public Health Genomics. Public Health Genomics 2009;12:121-128.



Delivering a **Healthy WA**



Government of Western Australia
Department of Health