RESEARCH SCHOOL OF BIOLOGY
DIRECTOR'S SEMINAR SERIES

THE AUSTRALIAN NATIONAL UNIVERSITY

PERSONALIZED INDIGENOUS GENOMES – DEFINING THE EXTENT OF MODERN HUMAN DIVERSITY

Dr. Vanessa Hayes
After graduating with a PhD in Cancer Genetics, University of Groningen, Netherlands (1999), heading a Laboratory investigating genetic susceptibility to HIV/AIDS in South Africa, Dr Hayes moved to Australia in 2003. As group leader of Cancer Genetics first at the Garvan Institute of Medical Research (2003 to 2008), she was recruited to the Children's Cancer Institute Australia and the new Lowy Cancer Research Centre (Sydney) in February 2008.

Dr Hayes has a keen interest in utilising state-of-the-art genetic-based technologies (including high-throughput genotyping platforms and next-generation sequencing) to advance her research into both human cancer and more recently a facial cancer threatening the imminent extinction of Australia's largest carnivorous marsupial, the Tasmanian devil.

1-2 PM THURSDAY 24 JUNE 2010

Venue: **The Robertson Lecture Theatre**
R.N. Robertson Building (no. 46)
Research School of Biology

Speaker: **Dr Vanessa M. Hayes**
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The era of personalized medicine foresees an increase in the field of personalized genomics. Before February 2010, there were only two named and published genomes, that of James Watson and Craig Venter. In this study we present the first complete indigenous personal genomes, that of the Archbishop Desmond Tutu, a Southern African Bantu (Niger-Congo B), and that of !Gubi, a Bushmen (Khoisan) from the Kalahari desert. Additional exome sequencing of three named Bushmen provides a glimpse into modern mans evolutionary past.

Indigenous peoples have predominantly been excluded from the efforts not only focused on personalized genomics, but also those of the Human Genome Project, the HapMap Project and the current 1000 Genomes Project. As a result, research efforts based on genetic disease association studies and drug responsiveness is largely limited to recently diverged populations, limiting global understanding of disease.

The Kalahari Bushmen represent one of the remaining hunter-gatherer societies, living for centuries along side agricultural Bantu-speaking peoples. We define in this study the most divergent genomes sequenced to date, with over 1.3 million newly described variants. These variants (common and rare, coding and non-coding) may not only reflect ancient adaptation to a foraging lifestyle, shed light into human transition and expansion as farmers, but importantly will contribute to a better understanding of current disease allele classifications and pharmacogenetics.

The novel variant content will be used to define genetic diversity across the diverse Southern African Bushman and Bantu populations. Our preliminary studies show the Bushmen represent an anciently divergent population, with as much genetic diversity between two Bushmen as between a European and an Asian. Defining the extent of genetic diversity within modern man will advance a global understanding of the human species and human disease.