

## Publication

### Patent

Human SNPs and Complex Physical Characteristics

### Journal Articles

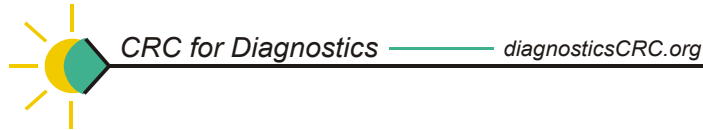
Voisey J., Box, N.F., and van Daal, A. (2001) A polymorphism study of the human agouti gene and its association with MC1R. *Pigment Cell Res.* **14**, 264-267.

Voisey, J. and van Daal, A. (2002) The role of Agouti Signal Protein in humans- OR - Agouti: From mouse to man, from skin to fat pigment. *Cell Res.* **15**, 10-18.

Voisey, J., Kelly, G., and van Daal, A. (2003) Agouti Signal Protein regulation in human melanoma cells. *Pigment Cell Res.* **16**, 65-71

## Information

Dr Angela van Daal  
Project Leader  
a.vandaal@qut.edu.au  
Tel: +61 7 3864 2502, Fax: +61 7 3864 1534



### Participants

Child Health Research Institute Inc.  
CSIRO  
La Trobe University  
Queensland Medical Laboratory  
Queensland University of Technology

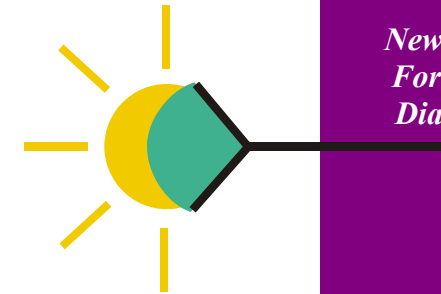
Mr Paul Barrett  
Business Development Director  
pg.barrett@qut.edu.au  
Tel: +61 7 3864 4297, Fax: +61 7 3864 5346

c/o QUT School of Life Sciences  
GPO Box 2434  
Brisbane QLD 4001  
AUSTRALIA  
Tel: +61 7 3864 1296  
Fax: +61 7 3864 5346  
E-mail: cdx@qut.edu.au  
Web: diagnosticsCRC.org



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## Human Identification Markers



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## Complex Human Characteristics



## Inspiration

Genetic variation at the level of single nucleotide polymorphisms (SNPs) is responsible for most genetic diversity between individuals. This accounts for differences in physical appearance (colouring, height, weight, etc.) and differences in susceptibility to disease (obesity, diabetes, heart disease, cancer).

The identification of individual SNPs underlying the heritable component of these complex human characteristics will allow them to be combined to form new diagnostic arrays. The elucidation of the underlying biological changes associated with these polygenic traits may lead to the development of novel therapeutic treatments.

## Investigation

### *Population Samples*

We are amassing a collection of DNA samples from individuals exhibiting a wide range of phenotypes (appearances) along with personal data for a number of complex traits such as pigmentation and height.

### *Novel SNPs and Data Mining*

We are screening candidate genes for novel SNPs in an effort to discover those SNPs associated with complex human characteristics. We also are using a genome-wide SNP scan to find SNPs associated with particular traits.

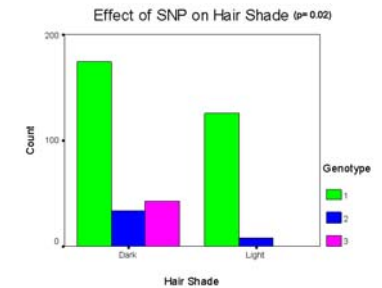
We are using many SNP databases to locate SNPs in candidate genes. We select those likely to affect function and look for associations with particular phenotypes.

## Application

SNPs can be combined to predict phenotypes. SNPs shown to affect function can be used as targets for therapeutics in conditions such as obesity.

## Validation

Preliminary studies already have identified a number of SNPs that show association with physical characteristics of pigmentation & obesity.



## Participation or Donation

*Would you like to take part in this cutting edge research? Perhaps you may wish to provide financial support for this project?*



Please contact Dr van Daal

Email [a.vandaal@qut.edu.au](mailto:a.vandaal@qut.edu.au) Tel (07) 3864 2502

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