

Title of Project:

Interactive Visual Analysis of SNP Data for Rapid Autozygosity Mapping in Consanguineous Families

Principle Investigator: Dr Ian M. Carr

Aim(s) of Project: To allow the rapid detection of autozygous regions linked to recessive disease genes in complex consanguineous pedigrees that can not be analysed by conventional methods. (This is a completed project see Hum Mutat 27(10), 1041– 1046, 2006).

Please provide a summary of the project proposal (max 1 page):

Autozygosity mapping of recessive disorders using small numbers of highly inbred families is a powerful tool for disease gene identification. However, the correct statistical analysis of these family pedigrees is not currently possible due to the very high computational demands involved. AutoSNPa is a computer program used for handling and visually presenting large amounts of SNP data, in such a way as to facilitate the rapid identification and subsequent scrutiny of autozygous regions. Hum Mutat 27(10), 1041–1046, 2006.

Please list 3 deliverables that the project will contribute to the UK and/or international cancer informatics community

The software allows the rapid identification of chromosomal regions 'Identical By Descent' in consanguineous families. In families that contain patients affected by a recessive condition (possibly predisposing to cancer), the defective gene will probably lay within one of these regions. Therefore the computer program identifies the probable location of disease causing genes in inbred pedigrees.

Please describe how the project will incorporate and/or reuse existing informatics infrastructure and/or resources. If the project will not use any existing infrastructure or resources (e.g. data standards or ontologies) please explain why this is the case.

This software program accepts data formatted as an Affymetrix results 'xls' file. Exported data is designed to be used by the researcher and is either an image file or a formatted plain text file.

Please describe the plans for the sharing of data and dissemination of knowledge that arise from the project:

This is a free to use computer program that can be downloaded from <http://xserve1.leeds.ac.uk/~iancarr/autosnpa/>.