iSNP – A Stratified Medicine Pipeline

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The human genome contains genetic variations, called Single Nucleotide Polymorphisms (SNPs) that underlie susceptibility to diseases such as diabetes and cancer. For inflammatory bowel disease (IBD), the severity and prognosis of the illness and the response to treatment have been linked to these genetic variations. Currently IBD costs the NHS £700 million annually, which is set to rise as this chronic disorder has a peak incidence in 3rd decade of life and are lifelong diseases.

'Omics technologies, which can identify and characterise every gene, have provided data, which combined with network biology expertise, can be utilised to assess the impact of individual SNPs on disease susceptibility and progression. However, a platform is needed to combine SNP genomics with advances in 'omics and network biology. Manually, this process can take four months to analyse one SNP, a faster, automated platform is therefore required.

Prof Simon Carding (IFR/UEA), Dr Tamas Korcsmaros (TGAC/IFR) and Dr Jo Brooks (IFR/UEA/NNUH) have developed and validated a workflow for SNPs in IBD with paradigm shifting potential. The aim of the translational project is to develop an automated version of the pipeline called iSNP – integrated SNP Network Pipeline. iSNP will effectively reduce the time and resources required to identify and assess the functional attributes of SNPs so the analysis can be automated and completed within one day. This means genetic mutations can be identified more than 100 times faster than is currently possible. This will dramatically reduce the cost of analysis of this data leading to improved patient benefit.