First Irish genome sequenced

September 7 2010

The first entire genome of an Irish individual has been sequenced. The sequence is reported in BioMed Central's open access journal, *Genome Biology* and provides insight into the evolutionary history of this distinct lineage.

Led by Professor Brendan Loftus, the research team from UCD Conway Institute used data from a previous genotyping study to select a suitable Irish male representative for sequencing. Then, using pair- and single-ended Illumina short read sequencing, one of the next generation sequencing approaches, the team created 9 DNA sequence libraries, which were overlaid to generate a high quality genome sequence with 11-fold coverage. Analyses were carried out in conjunction with collaborators from Trinity College Dublin, Royal College of Surgeons of Ireland (RCSI), Beaumont hospital, the MRC Human Genetics Unit and University of Edinburgh.

The researchers used HapMap and previous gene association studies to identify new DNA variants such as insertions/deletions (indels) and single nucleotide polymorphisms (SNPs).

Nearly 200,000 indels and over 3 million SNPs were identified in the Irish genome sequence. Of the SNPs, 13% were novel, potentially including markers specific to Irish ancestry or indicators of disease. In particular, one of the new SNPs interferes with the production of a macrophage-stimulating protein, thought to be associated with inflammatory bowel disease and chronic liver disease.

The authors also describe a new way to improve SNP calling accuracy at low genome coverage by using haplotype data from the current Human Genome Diversity Panel and they identify gene duplication events that may show recent positive selection in the human lineage.

"Our findings show that there remains utility in generating whole genome sequences to illustrate both general principles and reveal specific instances of human biology", says Loftus, adding, "The Irish population is of interest to biomedical researchers because of its isolated geography, ancestral impact on further populations and the high prevalence of a number of diseases".

The DNA variants in this study, funded through a Science Foundation Ireland Research Professorship award, have been uploaded to the Galaxy computing platform, facilitating the continued full analysis of this interesting genome. The authors hope that this sequence will compliment the ongoing 1000 genomes project, which currently lacks an Irish representative.

More information: Sequencing and analysis of an Irish human genome, Pin Tong, James GD Prendergast, Amanda J Lohan, Susan M Farrington, Simon Cronin, Nial Friel, Dan G Bradley, Orla Hardiman, Alex Evans, James F Wilson and Brendan J Loftus, *Genome Biology* (in press), genomebiology.com/

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