Docket #: S11-021

Annotation of Genome-Wide Genetic Variants Using a New Population-Specific Major Allele Reference

Stanford researchers have developed three novel human reference genome sequences, which will significantly improve the interpretation of the growing genetic data stemming from the human genome project and other related draft sequences. While these new sequences are based on the most common population-specific DNA sequence, they are unique because they are ethnicity-specific. This collection of major allele reference sequences provides a resource for ethnicity-aware interpretation of whole genome sequence data, providing a solution to inherent biases of the current reference sequence. They have captured most of the population-specific genetic variations present in three major population groups. The researchers also developed methods that integrate the ethnicity-concordant major allele reference sequences into genome interpretation pipelines.

Applications

- Ethnicity-aware mapping of massively parallel whole genome sequence data
- Identification of variation from the dominant ethnicity-specific DNA sequence for use in downstream genome interpretation pipeline applications

Advantages

- Works with existing short-read mapping and variant calling algorithms
- Improves accuracy of genetic sequence calling and mapping
- Provides a method for incorporating ethnic background into downstream genome sequence interpretation

Patents

- Published Application: 20130073217
- Issued: 10,127,346 (USA)

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