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A Method to Integrate Population and Familial Haplotype Phasing into Estimates of Genome-Wide Genetic or Gene Product Risk

Stanford researchers have developed a method for estimating the risk for disease on a genome-wide scale while incorporating population and familial haplotype phasing. Unlike current methods, this technology assigns the parental origin of genetic variants in interpreting the genome, improving the overall risk assessment of individual genomes. The assessment of compound heterozygous and multigenic disease risk, integration of sex-specific inheritance and integration of genetic background in areas of high recombination are all wholly reliant on phased haplotype data. This novel long-range haplotype phasing is based upon several databases, including family pedigree data, inheritance state determination, and population linkage disequilibrium data. The use of this technology not only improves risk prediction for personal genomes but also dramatically enhances the sensitivity of whole genome sequencing of related individuals for the identification of disease gene loci.

Applications

- Mapping of risk allele flow through extended pedigrees
- Performance of genotype-based determination of immunophenotypes such as HLA type using whole genome sequencing
- Integration of parent-of-origin information into risk prediction pipelines for interpretation of personal genomes from families
- Identification of multigenic and compound heterozygous contribution to inherited disease syndromes

Advantages

- Provides rapid long-range haplotype phased data
- Incorporates family pedigree information into pipelines for genome annotation
- Uses multiple data-sources rapid long-range haplotype phasing, with family pedigree information alone informing the vast majority of phased position

Patents

- Published Application: 20150370959

Innovators

- Frederick Dewey
- Jake Byrnes
- Carlos Bustamante
- Atul Butte
- Euan Ashley
- Rong Chen

Licensing Contact

Imelda Oropeza

Senior Licensing Manager, Physcial Sciences

Email