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A Method Incorporating Network Modeling to Enlarge the Search Space of Candidate Genes for Known Diseases (NetGBA)

Stanford researchers have developed a novel method that enlarges the search space for disease-gene relationships. The main barrier to genome interpretation is the inherent difficulty in prioritizing the millions of genetic variants in known genes. This technology improves upon currently available methods in several ways. First, it analyzes multiple data sources to prioritize genetic variants. It also utilizes curated disease-gene association to serve as a scaffold as it identifies and establishes disease genes. Furthermore, it uses network modeling to identify significant gene-gene associations for known diseases. Overall, this technology advances the ability to interpret genome sequences for personalized health care. It provides a novel solution for identifying candidate disease genes in individuals with inherited disease syndromes and recognizing risk alleles and carrier states in disease genes in healthy individuals.

Applications

- Mapping causal gene loci in inherited disease syndromes using whole genome sequence data
- Creating putative disease and carrier state predictions from whole genome sequencing
- Identifying targets for research on the pathophysiology of inherited disease syndromes
- Prioritization of genes for designing capture-based sequencing platforms and hybridization-based genotyping technologies for rapid molecular diagnosis of inherited disease syndromes

Advantages

- Incorporates multiple sources of prior information in defining gene-gene associations
- Enlarges the search space for disease genes to prioritize genetic variants using phenotypic information
- Greatly reduces the number of candidate variants in co-segregation studies of whole genome or exome sequence data for disease gene identification

Patents

- Published Application: 20130090908
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Innovators

- Frederick Dewey
- Euan Ashley

Licensing Contact

Imelda Oropeza

Senior Licensing Manager, Physical Sciences

Email