Docket #: S14-090

Sequence to medical phenotypes: a framework for identifying clinical phenotypes in whole genome DNA sequence data

Stanford inventors have developed a series of methods which can identify genetic variants and medical genotypes through the phasing of genetic data with the use of Mendelian inheritance as quality control. Additionally these methods allow for predictive genetic information regarding rare disease phenotype risk and responses to pharmacological therapy. This is brought about given the paradox of low price human genome sequencing with the difficulty of interpreting this information and being able to use it in a clinical context.

Applications

- Identification of medically actionable genetic variants with disease risk implications in pre-symptomatic individuals in WGS data
- Identification of genetic variants in WGS associated with drug response.
- Identification of genetic variants in WGS associated with disease in families and individuals

Advantages

- There does not yet exist, to date, a readily available pipeline for comprehensive clinical interpretation of sequence variants that integrates predicted methods and the bulk of published disease-genotype associations for genetic assessment of inherited disease risk and predicted drug response. This set of methods does just that.

Patents

- Published Application: 20150261913
- Published Application: 20200251178

Innovators

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