Docket #: S11-077B

A Method and System to Phase an Individual Genome With or Without Family Members in the Context of Clinical Interpretation-B

The present disclosure presents a unified system to phase a personal genome for downstream clinical interpretation. In an embodiment, an initial phasing is generated using public datasets, such as haplotypes from the 1000 Genomes Project, and a phasing toolkit. A local perturbation algorithm is applied to improve long range phasing. If available, a Mendelian inheritance pipeline is applied to identify phasing of novel and rare variants. These datasets are merged, followed by correction by any experimental data. This allows for full clinical interpretation of the role of a group of variants in a gene, whether inherited or de novo variants.

Applications

- Interpretation of a personal genome
- Full clinical ointerpretation of the role of a group of gene variants

Advantages

- Improved long range phasing
- Applied Mendelian inheritance pipeline if available

Patents

- Published Application: 20130085728

- Issued: 9,928,338 (USA)

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