

SYSTEMS AND METHODS FOR VISUALIZING STRUCTURAL VARIATION AND PHASING INFORMATION

ABSTRACT

A system for providing structural variation or phasing information is provided. The system accesses a nucleic acid sequence dataset corresponding to a target nucleic acid in a sample. The dataset comprises a header, synopsis, and data section. The data section comprises a plurality of sequencing reads. Each sequencing read comprises a first portion corresponding to a subset of the target nucleic acid and a second portion that encodes an identifier for the sequencing read from a plurality of identifiers. One or more programs in the memory of the system use a microprocessor of the system to provide a haplotype visualization tool that receives a request for structural variation or phasing information from the dataset. The request is evaluated against the synopsis thereby identifying portions of the data section. Structural variation or phasing information is formatted for display in the haplotype visualization tool using the identified portions of the data section.

FIG. 1