Unified detection, classification, and phasing of germline, somatic, and denovo variation from high throughput sequencing data

Daniel Cooke and Gerton Lunter

August 5, 2016

Abstract

High throughput sequencing technology has seen the rise of powerful methods to detect genetic variation directly from raw data. Most notibly, haplotype-based variant calling has become defacto for identifying germline variation.

Motivation

Methods

0.0.1 Read preprocessing

0.0.2 Candidate variant generation

0.0.3 Candidate haplotype generation

0.0.4 Model fitting

0.0.5 Calling and refinement

Results

Discussion