Identification of Germline Mutations in Familial Syndromes Using Sequencing Data

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Abstract

Predicting carrier probability of germline mutations in known susceptibility genes is becoming common practice in cancer prevention. Various studies have shown that accounting for Mendelian transmission of germline mutations using family history data can significantly improve prediction accuracy. Recently, similar methods are proposed to improve the identification of unknown germline variants in sequencing studies of family trios. Here, we describe a new method that performs joint variant calling using raw sequencing measurements, i.e., intensities or counts, which are generated from related family members. Our method is applicable to families with extended pedigree structures. It is most useful for the identification of new disease genes that harbor mutations with minor allele frequency (MAF) < 0.1%.

Keywords: DNA sequencing; Rare variants.

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