

عنوان مقاله:

Beyond genome wide association study: steps through precision medicine in Tehran cardio-metabolic genetic study

محل انتشار:

سومین کنگره بین المللی پزشکی شخصی ایران (سال: 1397)

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خلاصه مقاله:

Precision medicine strives to delineate disease using multiple data sources—from genomics to digital health metrics—in order to be more precise and accurate in our diagnoses, definitions, and treatments of disease subtypes. By defining disease at a deeper level, we can treat patients based on an understanding of the molecular underpinnings of their presentations, rather than grouping patients into broad categories with one-size-fits-all treatments. The primary goal of GWAS is to detect associations between genetic variants and traits in samples from populations in order to better understand the biology of disease, under the assumption that a better understanding will lead to prevention or better treatment. The path from GWAS to biology is not straightforward because an association between a genetic variant at a genomic locus and a trait is not directly informative with respect to the target gene or the mechanism whereby the variant is associated with phenotypic differences. Findings from GWAS could be used as some selected markers in precision medicine. Tehran cardio-metabolic genetic study used deep whole genome sequencing data besides chip typed information to impute more than 60 million genetic markers on more than 15000 Iranian individual to build a unique and well phenotype dataset. This dataset is a good infrastructure for doing scientific research in the field of precision medicine in Iranian population.

کلمات کلیدی:

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