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Editorial

Genome-Wide Association Study (GWAS): Methodology and Applications

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INTRODUCTION

A genome-wide association study (GWAS) is a method of genetic analysis that uses linking certain genetic variants to specific diseases. The process entails scanning the genomes of a large number of people in order to identify genetic markers that can be used to predict the presence of a disease. The frequency and distribution of SNPs are often compared between individuals with an illness or trait and those without a disease or trait in GWASs. The threshold for significance in a GWAS is quite high due to multiple testing, which involves analysing hundreds of thousands of SNPs (corresponding to a very small P value).

Genome-wide association studies (GWAS) are a frequent study strategy for determining connections between common DNA sequence variants and human attributes. GWA studies evaluate the DNA of individuals with varied phenotypes for a certain trait or disease when applied to human data.These individuals could be people who have a disease (cases) and people who do not have the disease (controls), or they could be people who have different phenotypes for a specific variable, such as blood pressure. The multiple statistical analyses run on GWAS data must be assessed with an inflated rate of false positives in mind. Systematic stratifications that can exist across thousands of genetic parameters, most notably due to disparities in the genetic heritage of research participants, require further modifications.

In contrast to approaches that evaluate a restricted number of pre-specified genomic areas, GWA research look at the complete genome.In contrast to gene-specific candidate-driven investigations, GWAS is a non-candidate-driven technique.GWA research can find SNPs and other DNA variants linked to an illness, but they can't tell you which genes are to blame.Results from several GWAS can be merged using other techniques to increase the ability to find SNPs with very tiny impact on the phenotype. While the GWAS design hasn't improved the prediction of prevalent illness features significantly, it has led in thousands of new connections between DNA base-pair alterations and the qualities they influence.

Myocardial infarction was the subject of the first successful GWAS, which was published in 2002.This study approach was later used in the landmark GWA 2005 study, which looked at individuals with age-related macular degeneration and discovered two SNPs with significantly different allele frequencies than healthy controls.The identification of causal genetic variations and subsequent characterization of complex genetic features has been revolutionised by genome-wide association studies (GWAS).GWAS is a powerful tool for identifying rare variants that contribute just a small amount to the overall trait heritability of disorders with complicated symptoms, such as polycystic ovary syndrome and premature ovarian insufficiency/failure.

The "common disease–common variation" concept states that genetic risk is determined by shared sequences of DNA carried with variable frequencies in cases and controls and inherited from ancient ancestors.The application of the findings in a way that accelerates drug and diagnostics development, such as better integration of genetic studies into the drug-development process and a focus on the role of genetic variation in maintaining health as a blueprint for designing new drugs and diagnostics, is a challenge for future successful GWA studies.

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