Research and Reviews: Drug Delivery

Recent Applications of Genome Wide Association Studies

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Perspective

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ABOUT THE STUDY

GWAS has been widely utilised to discover novel genes that contribute to the development of Parkinson's disease. Huge sample sizes are required, and the data set's genotypes are used to compare persons with PD to an equally large group of control subjects. There were 13,708 cases and 95,282 controls in a big meta-analysis that used numerous previously published individual GWAS searches. This study confirmed 24 previously identified Single Nucleotide Polymorphisms (SNPs) and discovered six new ones. An intriguing technique of displaying the known genes linked in Parkinson's disease. This included 6476 PD cases of European ancestry who were not related to those previously studied in (2014), as well as 302,042 controls

that did not have PD but shared the same ancestry as the cases. The researchers discovered 17 new loci by integrating the data sets from these two studies.

The recent use of GWAS to liver disease research exemplifies how "discovery" GWAS studies can propel a topic that had been stagnant for almost a decade forward. NAFLD (Non-Alcoholic Fatty Liver Disease) is a complex disease that clusters in families and leads to cirrhosis and liver failure. Obesity and diabetes are the most common risk factors. Due to our lack of understanding of the disease's molecular foundation, there are no recognised therapeutic medications for NAFLD. Surprisingly, a single *PNPLA3* gene mutation was discovered to be strongly linked to the development of steatosis and NAFLD. *PNPLA3* encodes a 481-amino-acid patatin-like phospholipase protein with unclear function, and the cytosine to guanine substitution leads in an amino acid alteration at codon 148.

A follow-up research of 7,176 patients confirmed the link between *PNPL43* and NAFLD susceptibility and discovered variations in three additional loci linked to increased CT hepatic steatosis as well as histologic NAFLD. Because of the pharmacogenomics elucidation of the molecular targets implicated in the development of NAFLD, these findings are anticipated to pave the way for the development of new and effective therapeutics for the condition. Another example is the empirical creation of the first effective medicines for chronic hepatitis C infection, which was based on the known antiviral activity of human type I interferons, which are produced by leukocytes and involve the innate immune response.

For individuals with HCV genotype 2 or 3 infections, pegylated interferon-alpha plus ribavirin is currently the first-line treatment. However, sustained virology response rates to peginterferon-based therapy, which are only 50% in patients with genotype I HCV infection, have remained unchanged for over a decade, until the recent introduction of HCV protease inhibitors, a new class of therapeutic medicines for this disease. Despite extensive study, limited understanding of the molecular reasons driving huge variances and resistance in patient response to peg interferon-based medicines existed until the effective use of GWAS, which allowed therapy to be tailored to the individual.

Surprisingly, genetic variations near the human *IL28B* gene, which encodes for interferon lambda 3, were linked to significant disparities in treatment response and spontaneous clearance of genotype I hepatitis C virus. The discovery of "newer" types of non-SNP genetic variants, such as indels, CNVs, and copy-neutral variations (inversions and translocations), has lately broadened the scope of study to find genetic markers of human diseases and treatment response. The 1000 Genomes Project, a global partnership that built on the data and technology developed by earlier "big science" efforts, was established to create the most detailed map of genetic variations in the human genome by sequencing the genomes of at least 1,000 people from around the world.