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Understanding Genetic Epidemiology: Benefits and Challenges of Genetics in Human Health

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In an era of escalating healthcare expenses and disease burden, understanding the distribution and drivers of human disease is becoming increasingly important. Genetic epidemiology, which analyses the role of inherited variables in illness aetiology, sits at the intersection of genetics and epidemiology. The current public health benefits of genomics research include a better understanding of disease causes [1], targeted cancer therapy [2], and medication dose regimes [3]. To fully appreciate the current and potential future contributions of genetic epidemiology research to improving human health, it is necessary to first understand the major milestones in genetic epidemiology as well as the current challenges researchers face in the ongoing process of deciphering the human genome.

Genetic epidemiology is a relatively recent field that was initially outlined in 1954 by Neel and Schull [4, 5]. Molecular genetics was still in its infancy at the time. Although direct genotype measurement was not possible at the time, genetic epidemiologists looked into disease inheritance by comparing inheritance patterns (dominant, recessive, X-linked) to phenotype patterns seen in big families. Early segregation analyses of breast and ovarian cancer, for example, suggested a strong genetic aetiology with an autosomal dominant inheritance style [6, 7].

The paradigm for linkage analysis was initially described in 1980 [8], and it was a natural extension of segregation analysis. Linkage analysis used populations of related individuals to assess the genetic basis of disease and successfully identified the genes responsible for numerous monogenic disorders, including Tay-Sachs, Huntington's, and cystic fibrosis [9]. It was made possible by technological advances that allowed direct measurement of genotypes. Linkage analysis, on the other hand, was unable to discover genes linked to complex, chronic disorders in the general population. Genes linked to rare familial forms of breast cancer in certain families, such as polymorphisms in the GPT and ACP genes, were found to have no connection with breast cancer in the general population [10].

Due to the limitations of linkage analysis, researchers looked into different methods for identifying genes linked to complicated disorders, such as candidate gene studies. Candidate gene studies were popular because they provided increased power for discovering connections for complex variables and could be undertaken in population-based cohort and case-control studies [11]. Few candidate gene studies have been successful in identifying connections despite using biologic knowledge. In a 2009 evaluation of candidate gene studies for obesity, for example, only nine genes were identified to have any connection with obesity out of 21 genes evaluated in the candidate gene literature [12]. Furthermore, only a few of the positive findings were repeated in future investigations [13, 14].

Parallel to the surge in popularity of candidate gene research was the Human Genome Project's (HGP) publication of the first draught of the human genome in 2001 [15], which has had a long-term impact on the field of genetic epidemiology. In a nutshell, the HGP aimed to catalogue human genetic variation by identifying all human genes and sequencing the human genome's three billion bases [16]. Large-scale human genome studies, such as genome-wide association studies (GWAS), were made possible by the HGP, allowing researchers to evaluate connections between features of interest and single base pair variations called single nucleotide polymorphisms (SNPs) spread throughout the human genome [17]. On a large scale Candidate gene studies, which normally tested a small number of SNPs at a handful of pre-specified genes, were considerably expanded by GWAS. Genetic epidemiology has made great progress in understanding the genomic aetiology of complex diseases because to genome-wide association studies (GWAS). GWAS have effectively reported about 11,000 SNPs related with a wide array of illnesses and their risk factors as of August 2013 [18].

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Conflict of Interest

None

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