Why Do Association Studies?

Genetic epidemiology was first described by Neel and Schull in 1954, but it was defined by Morton, Chung and Mi (1967) as a "science that deals with etiology, distribution and control of disease in groups of relatives and with inherited causes of disease in populations."[1] It focuses on the genetic determinants of disease and the joint effects of genes and non-genetic determinants.[2] One of the main objectives of genetic epidemiology research is to identify genetic variants in disease susceptibility genes that may be responsible for the variable risk among individuals.[3]

Through the years, advances in science and development of new methodologies have facilitated for a greater appreciation of genetic epidemiology. The advent of whole genome sequencing and whole exome sequencing has allowed researchers to explore the full spectrum of genetic variation.[4]

Barbujani et al., found that genetic variation remains high even within small groups and at least four fifths of human genetic variation reflect individual differences.[5] Gene polymorphisms are the most common type of genetic variation in humans and are important contributors to interindividual variation and can result in both distinct phenotypes and genotypes.[6,7]

Studies that deal with variants are important in ensuring that populations are well represented especially in genome wide association studies (GWAS). This information contributes to the body of knowledge that is out there. Based on the GWAS catalog, 80% of current participants as being of European ancestry even though they only represent 16% of the whole populations,[8] which suggests that the data available is skewed towards a particular race. Thus, it is essential that other populations be included, and researchers play an active role to ensure diversity in research study populations.[9,10]

Studies can have the potential to make contributions to our understanding of polymorphisms and disease risks, and to educate scientists, physicians, public health professionals, and policy makers.[7]

In our country, there is more work to be done in terms of doing further research on association studies and variants. We have brilliant scientists and many conditions that have not been the subject of these types of research. It is only a matter of time before our population becomes well-represented in the genomic world.

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