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Challenges with gene-environment interactions: Where we are and where we need to go

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ABSTRACT

Environmental diseases are seen as the result of exposure to environmental stressors modulated by individual susceptibility factors. Human population studies typically involve measuring numerous biomarkers of both phenotype and genotype. Biomarkers can reflect exposure, effect (disease risk), or individual susceptibility. For environmentally-induced diseases, molecular biomarkers play a key role in understanding the relationships between environmental stressors and the development of chronic diseases, and in identifying individuals at increased risk.

The molecular epidemiological approach using molecular markers including single nucleotide polymorphisms (SNPs) has been in use for over a quarter of a century in the fields of environmental medicine, genetic epidemiology, and especially in monitoring environmental and occupational exposure, showing that genetic susceptibility to suspected chemical and environmental carcinogens may modify the response to exposure.

More recently, genome-wide association studies (GWAS) have offered various techniques to study DNA variations which are associated with human diseases. However, knowledge of the effect of SNPs on common diseases is still needed in order to understand the underlying genetic factors affecting human diseases – whether its gene-gene or gene-environment interactions.

Applying the prevailing 'omics' approaches (transcriptomics, metabolomics, exposomics, whateveromics) to study these interactions produces a superabundance of data, and as a result an urgent need for appropriate statistical and computational methods. Traditional statistical methods are inadequate, due to the high dimensionality of the data and the occurrence of multiple polymorphisms.

The future clearly lies with omics; but while a large number of genetic variants and gene-environment interactions have been explored for cancer and other environmental diseases, to date replication of studies is lacking and therefore the findings remain to be validated. There is still scope for novel standardized high throughput methodologies, more focused predictive biomarker assays based on specific endpoints, larger samples and cohorts, and better environmental assessment. The new multi-dimensional approaches provide great opportunities for prospective studies to identify and characterize the genes that interact with other genes and environment factors, and to elucidate their causative and modulatory influences on complex multifactorial diseases.