From genetic epidemiology to exposome and systems epidemiology

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Genetic epidemiology investigates the contribution of germline gene variants to disease etiology. The genetic architecture of many diseases exhibiting Mendelian inheritance patterns was decoded through family-based studies and subsequent functional cloning. Tests for high-penetrance gene variants have clinical utility for individuals, such as in preimplantation, prenatal, or postnatal diagnostics, and in preventive cascade screening of biological relatives. Genome-wide association studies thousands of identified variants associated with complex traits. Yet, the causal variants explaining the replicated statistical associations and their functional effects have rarely been deciphered. A missing heritability gap remains. Genetic testing for complex phenotypes has utility for research and public health rather than individuals. Genetics integrated with other biomarkers and information in the context of large citizen cohorts enables systems epidemiology approaches. The utility of genomic biomarkers is in studying molecular pathways mediating effects of the exposome on the phenome in order to improve causal understanding of modifiable risk factors. The goal is to strengthen the primary prevention of noncommunicable diseases.

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