



Fundamentals of Genetic Epidemiology

Muin J. Khoury, Terri H. Beaty, Bernice H. Cohen

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With continued progress in mapping and sequencing of the human genome, and increasing recognition of the role of genes in disease etiology, there is a need for a more sophisticated approach to the investigation of the causes of complex chronic diseases. This text integrates the principles, methods and approaches of epidemiology and genetics in the study of disease etiology. After a brief historical overview of genetics and epidemiology and their gradual rapprochement, the authors define the central theme of genetic epidemiology as the study of the role of genetic factors and their interaction with environmental factors in the occurrence of disease in populations. They describe fundamental research strategies of genetic epidemiology including population and family studies. Among the former are the study of the distribution of genetic traits and the role of nonspecific genetic indicators (such as inbreeding and admixture) in the occurrence of diseases. Among the latter are the analysis of familial aggregation of disease and its causes by epidemiologic methods as well as techniques of formal genetic analysis (variance components, segregation and linkage analysis). Finally, the authors discuss the increasing applications of genetic epidemiology in preventive medicine, public health surveillance, and the emerging ethical issues regarding use of genetic information in society.

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