Genomic data and epidemiological studies

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Epidemiology aims to understand the etiology of diseases. Genetics can contribute to this understanding in several ways. A few diseases are caused by a mutation or chromosome aberration, but the genetic contribution is usually more subtle. One way that genes affect disease is by modifying the impact of another risk factor. For example, the carcinogenic effect of a chemical dose may depend upon metabolic rates, which in turn depend on the genotypes of the enzymes in the metabolic pathway. Another way that genes affect disease is by modifying the effect of interventions. If the intervention is a drug, genetic variation in enzymes responsible for its metabolism may be the key event; conceptually a version of the previous example. For diseases like various cancers where somatic mutations are believed to play an important role in their progression, gene expression profiles are being explored as a way to divide patients into subsets that are more likely to benefit from an intervention. Such genetic considerations often underlie research portrayed as personalized nutrition / medicine. In this talk, two paradigms for discovering genetic contributions are discussed through examples from my research. One examines a statistical genetics problem in identifying genetic variants which affect metabolic rates. The other example discusses some of the many statistical challenges in using gene expression profiles to target intervention.