Statistical Problems of Genetic Mapping

David O. Siegmund

Stanford University

The goal of linkage analysis is to locate genes affecting particular traits (e.g., genes that affect human susceptibility to particular diseases or genes that affect productivity of agriculturally important species) by comparing the phenotypes and genotypes of related individuals. I will formulate linkage analysis of both qualitative and quantitative traits in humans and in experimental genetics as a change-point like problem and discuss from that viewpoint three statistical problems arising from genome scans to detect anonymous genes: (i) multiple comparisons due to simultaneous testing of many markers for linkage to the trait of interest; (ii) statistical power to map genes as a function of the true and the assumed genetic models, especially when there are multivariate phenotypes, gene-gene or gene-environment interactions; and (iii) confidence bounds for estimation of genetic effects.