专论与综述

人类复杂疾病关联研究中群体分层的检出和校正

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摘要

病例对照研究是鉴定多基因疾病易感位点重要的遗传流行病学方法，而群体分层是导致病例对照研究关联研究结果出现偏倚甚至是假关联的重要原因之一。文章对人群分层的检出及校正的方法和原理进行了阐述，包括基于核心家系的传递/不平衡检验(TDT)以及基于不相关基因组遗传标记的基因组对照(GC)和结构化关联(SA)等，并且对这几种方法进行了比较。

Detection and controlling for population stratification in association studies of human complex disease

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Abstract

<P>Case-control studies, which serve as standard design for genetic association analysis, can be the most practical and powerful approach to detect genetic polymorphisms contributing to susceptibility to complex human diseases. However, considerable concern has been expressed that this approach is prone to population stratification, which can lead to biased or spurious results. We review several methods to detect and account for population stratification; these methods include nuclear family-based transmission/disequilibrium test (TDT), and genomic control (GC) and structured association (SA) based on unlinked genetic markers.</P>

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