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肺结核全基因组关联研究进展

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摘要 肺结核是由结核分枝杆菌感染引起的一类古老但仍对人类造成巨大影响的传染性疾病。到目前为止, 肺结核依然是由单一病原菌导致死亡人数最多的疾病, 并且随着耐药菌株的出现而呈现死灰复燃之势。近几年, 肺结核全基因组关联研究在世界范围内取得了阶段性成果, 发现了与肺结核相关联的遗传易感位点和区域, 使肺结核的遗传学研究进入了一个崭新的阶段, 为后续肺结核的早期和综合防治提供了重要线索。然而, 由于人群遗传结构差异和宿主/病原体相互作用, 与其他复杂疾病相比, 肺结核全基因组关联研究依旧面临重重困难, 进展缓慢。文章对不同人群肺结核全基因组关联研究及其验证进行综述, 并系统阐述了目前研究中存在的困难及可能的应对策略。

关键词: 肺结核 全基因组关联研究 易感基因

Abstract: Tuberculosis, caused by Mycobacterium tuberculosis (MTB), is one of the oldest and most influential diseases in the history due to its devastating effect on health and high mortality rate worldwide. Tuberculosis causes more human deaths than any other single infectious disease and the incidence of the tuberculosis is increasing dramatically in recent years. Genome-wide association study (GWAS) has been used to delineate the genetic basis of tuberculosis, and several susceptibility genes and loci were found, which provids important clues to the early intervention and treatment of tuberculosis. However, due to difference in the population structure and host-pathogen interactions, GWAS on tuberculosis faces great challenges. In this review, we introduced the achievements of GWAS on tuberculosis, and illustrated challenges and strategies in the future study.

Keywords: tuberculosis, genome-wide association study, susceptibility gene

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