

本期目录 | 下期目录 | 过刊浏览 | 高级检索

[打印本页] [关闭]

## 综述

### DNA混合分析技术的单体型频率估计方法

何柳, 唐迅, 胡永华

北京大学医学部公共卫生学院流行病与卫生统计学系, 流行病学教育部重点实验室, 北京 100191

摘要:

DNA混合分析技术的广泛使用, 能够明显降低研究的工作量和成本, 然而仍然有其自身局限, 例如损失了大量个体遗传信息、测量误差较大等。为了尽量克服DNA混合分析技术的自身问题, 其检测和分析方法在不断发展与完善。在单体型频率估计方面, 基于最大期望(expectation-maximization, EM)算法的新方法不断涌现, 如HaploPool算法和PoooL算法, 其准确性、实用性均增强。

关键词: DNA混合分析技术 单体型 遗传流行病学

### Estimation of haplotypes based on DNA pooling

HE Liu, TANG Xun, HU Yonghua

Department of Epidemiology & Biostatistics, Key Laboratory of Epidemiology of Ministry of Education, Health Science Center, Peking University, Beijing 100191, China

Abstract:

DNA pooling, a fast and economic study strategy, is widely used in areas of scientific research. In spite of various limits, researchers are making their efforts to improve DNA pooling toward a more perfect direction, including allele frequency detection and estimation of haplotypes. In haplotype estimation, more and more analyzing methods originated from the expectation-maximization algorithm have appeared, with improved accuracy and practicality, such as HaploPool algorithm and PoooL algorithm.

Keywords: DNA pooling haplotypes genetic epidemiology

收稿日期 2010-04-22 修回日期 网络版发布日期

DOI: 10.3969/j.issn.1672-7347.2011.

基金项目:

国家自然科学基金(30671807, 30872173)。

通讯作者: 胡永华,E-mail:yhhu@bjmu.edu.cn

作者简介: 何柳, 博士研究生, 主要从事遗传流行病学方面的研究。

作者Email: yhhu@bjmu.edu.cn

## 参考文献:

- [1] Arnheim N C, Strange H E. Use of pooled DNA samples to detect linkage disequilibrium of polymorphic restriction fragments and human disease: Studies of HLA class II loci [J]. Proceedings Nat Acad Sci, 1985, 82(20):6970-6974.
- [2] Bharti A R, Letendre S L, Patra K P, et al. Malaria diagnosis by a polymerase chain reaction-based assay using a pooling strategy [J]. Am J Trop Med Hyg, 2009, 81(5):754-757.
- [3] Bugeja M J, Booth D, Bennetts B, et al. An investigation of polymorphisms in the 17q11.2-12 CC chemokine gene cluster for association with multiple sclerosis in Australians [J]. BMC Med Genet, 2006, 7: 64.
- [4] Matkovich S J, Van Booven D J, Hindes A, et al. Cardiac signaling genes exhibit unexpected sequence diversity in sporadic cardiomyopathy, revealing HSPB7 polymorphisms associated with disease [J]. J Clin Invest, 2010, 120(1): 280-289.
- [5] Baro J A, Carleos C, Corral N, et al. Power analysis of QTL detection in half-sib families using selective DNA pooling [J]. Genet Sel Evol, 2001, 33(3):231-247.
- [6] Sham P, Bader J S, Craig I, et al. DNA Pooling: a tool for large-scale association studies [J]. Nat Rev Genet, 2002, 3(11):862-871.

扩展功能

本文信息

► Supporting info

► PDF(898KB)

► [HTML全文]

► 参考文献[PDF]

► 参考文献

服务与反馈

► 把本文推荐给朋友

► 加入我的书架

► 加入引用管理器

► 引用本文

► Email Alert

► 文章反馈

► 浏览反馈信息

本文关键词相关文章

► DNA混合分析技术

► 单体型

► 遗传流行病学

本文作者相关文章

PubMed

DNA混合分析技术的单体型频率估计方法 何柳, 等封三 [7] Pearson J V, Huentelman M J, Halperin R F, et al. Identification of the genetic basis for complex disorders by use of pooling-based genomewide single-nucleotide-polymorphism association studies [J]. Am J Hum Genet, 2007,80(1):126-139.

[8] De Bakker P I, Graham R R, Altshuler D, et al. Transferability of tag SNPs to capture common genetic variation in DNA repair genes across multiple populations [J]. Pac Symp Biocomput, 2006,11:478-486.

[9] International HapMap Consortium. The International HapMap Project [J]. Nature, 2003,426(6968):789-796.

[10] Hoh J, Matsuda F, Peng X, et al. SNP haplotype tagging from DNA pools of two individuals [J].

BMC Bioinformatics, 2003, 4: 14.

[11] Ito T, Chiku S, Inoue E, et al. Estimation of haplotype frequencies, linkage-disequilibrium measures, and combination of haplotype copies in each pool by use of pooled DNA data [J]. Am J Hum Genet, 2003,72(2):384-398.

[12] Yang Y, Zhang J, Hoh J, et al. Efficiency of single-nucleotide polymorphism haplotype estimation from pooled DNA [J]. Proc Natl Acad Sci U S A, 2003,100(12):7225-7230.

[13] Barrett J C, Fry B, Maller J, et al. Haplovview: analysis and visualization of LD and haplotype maps [J]. Bioinformatics, 2005,21(2):263-265.

[14] Kirkpatrick B, Armendariz C S, Karp R M, et al. HaploPool: improving haplotype frequency estimation through DNA pools and phylogenetic modeling [J]. Bioinformatics, 2007,23(22):3048-3055.

[15] Philip E B. RECOMB' 04: Proceedings of the Eighth annual international Conference on Resaerch in Computational Molecular Biology [M]. New York,USA: ACM Press,2004: 10-19.

[16] Stephens M, Smith N J, Donnelly P. A new statistical method for haplotype reconstruction from population data [J]. Am J Hum Genet, 2001,68(4):978-989.

[17] Zhang H, Yang H C, Yang Y. PoooL: an efficient method for estimating haplotype frequencies from large DNA pools [J]. Bioinformatics, 2008,24(17):1942-1948.

[18] Zhang H, Zhang H, Li Z, et al. Statistical methods for haplotype-based matched case-control association studies [J]. Genet Epidemiol, 2007,31(4):316-326.

[19] Valle T, Tuomilehto J, Bergman R N, et al. Mapping genes for NIDDM. Design of the Finland-United States investigation of NIDDM Genetics (FUSION) study [J]. Diabetes Care, 1998,21(6):949-958.

[20] Lin D Y, Zeng D. Likelihood-based inference on haplotype effects in genetic association studies [J]. J Am Stat Assoc, 2006,101(473): 89-104.

[21] Yang Y, Hoh J, Xu F, et al. Efficiency of SNP haplotype estimation from pooled DNA [J].

Proceedings Nat Acad Sci, 2003,100(12):7225-7230.

[22] Jain S, Tang X, Narayanan C S, et al. Angiotensinogen gene polymorphism at -217 affects basal promoter activity and is associated with hypertension in African-Americans [J]. J Biol Chem, 2002, 277(39): 36889-36896.

[23] Yang H C, Pan C C, Lin C Y, et al. PDA: Pooled DNA analyzer [J]. BMC Bioinformatics, 2006,7:233.

[24] Niu T, Qin Z S, Xu X, et al. Bayesian haplotype inference for multiple linked single-nucleotide polymorphisms [J]. Am J Hum Genet, 2002,70(1):157-169.

[25] Zuo Y, Zou G, Zhao H. Two-stage designs in case-control association analysis [J]. Genetics, 2006, 173(3):1747-1760.

本刊中的类似文章