中南大学学报(医学版) 2010, 35(9) 915- DOI: 10.3969/j.issn.1672-7347.2010. ISSN: 1672-7347 CN: 43-1427/R

论著

PTP1B基因多态性与儿童单纯性肥胖的关系

莫娟， 吴静， 孙志香， 杨浩波， 雷闽湘， 刘玮芳

中南大学湘雅医院内分泌科， 长沙 410008

摘要：

目的：分析儿童蛋白酪氨酸磷酸酶1B(PTP1B)基因IVS6+G82A与Pro303Pro多态性分布特征，探讨IVS6+G82A与Pro303Pro遗传多态性在儿童单纯性肥胖发病机制中的作用。 方法：随机抽取147例肥胖儿童及118例健康儿童，应用聚合酶链反应-限制性片段长度多态性(polymerase chain reaction-restriction fragment length polymorphism, PCR-RFLP)方法检测PTP1B基因IVS6+G82A与Pro303Pro多态性。同时检测腰围(waist circumference, WC)、腰臀比(waist to hip ratio, WHR)、体脂百分比(percentage of body fat, %BF)、收缩压(systolic blood pressure, SBP)、舒张压(diastolic blood pressure, DBP)、空腹血糖(fasting plasma glucose, FPG)、血清甘油三酯(serum triglycerides, TG)、总胆固醇(total cholesterol, TC)、高密度脂蛋白胆固醇(high density lipoprotein-cholesterol, HDL-C)、低密度脂蛋白胆固醇(low density lipoprotein-cholesterol, LDL-C)、空腹胰岛素水平(plasma fasting insulin, FINS)、稳态模型胰岛素抵抗指数(homeostasis model assessment for insulin resistance, HOMA-IR)和瘦素水平。 结果：PTP1B基因IVS6+G82A与Pro303Pro多态性发生频率健康儿童分别为53.4%与11.0%，肥胖儿童分别为59.5%与19.4%。肥胖儿童PTP1B基因IVS6+G82A多态性与健康儿童差异无统计学意义，且该位点突变与肥胖儿童临床指标无明显相关性。肥胖儿童PTP1B基因Pro303Pro多态性与健康儿童相比，基因型分布与等位基因频率均差异有统计学意义，并且其多态性与体质量指数, WC, TG和LDL-C水平有关。IVS6+G82A与Pro303Pro连锁不平衡分析显示两位点间连锁不平衡很弱(D′: 0.441, r2: 0.027)。 结论：PTP1B基因IVS6+G82A变异与儿童单纯性肥胖无明显相关性，而Pro303Pro变异可能与儿童单纯性肥胖相关，且影响肥胖儿童的脂质代谢。

关键词： 儿童 肥胖 蛋白酪氨酸磷酸酶1B 单核苷酸多态性 瘦素 胰岛素

Association of PTP1B gene polymorphism with obesity in Chinese children

MO Juan, WU Jing, SUN Zhixiang, YANG Haobo, LEI Minxiang, LIU Weifang

Department of Endocrinology, Xiangya Hospital, Central South University, Changsha 410008, China

Abstract:

ObjectiveTo investigate the distribution characteristics of protein tyrosine phosphatase 1B (PTP1B) gene IVS6+G82A and Pro303Pro polymorphisms in Chinese children and determine the effect of PTP1B gene IVS6+G82A and Pro303Pro polymorphisms on the pathogenesis of childhood obesity. MethodsA total of 147 Chinese obese and 118 healthy children were randomly selected and enrolled to identify IVS6+G82A and Pro303Pro genotypes by polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) assay. Waist circumference (WC), waist to hip ratio (WHR), percentage of body fat (%BF)，systolic blood pressure (SBP), diastolic blood pressure (DBP), fasting plasma glucose (FPG), serum triglycerides (TG), total cholesterol (TC), high density lipoprotein-cholesterol (HDL-C), low density lipoprotein-cholesterol (LDL-C), plasma fasting insulin (FINS), homeostasis model assessment for insulin resistance (HOMA-IR), and plasma leptin were examined. ResultsThe allele frequencies of IVS6+G82A and Pro303Pro were 59.5% and 19.4% in obese children, and 53.4% and 11.0% in healthy children, respectively. There were significant differences in allele frequencies of Pro303Pro polymorphism between the obese and the control group. Pro303Pro polymorphism was associated with body mass index, WC, TG, and LDL-C in the obese subjects. There was not di-fference in the genotype distributions or allele frequencies of IVS6+G82A polymorphism between the obese and the control group. Further analysis showed no association between the genotypes of IVS6+G82A and clinical characteristics in the obese subjects. The linkage disequilibrium analysis for IVS6+G82A and Pro303Pro (D′: 0.441, r2: 0.027) was weak.ConclusionPTP1B gene Pro303Pro polymorphism might be associated with the pathogenesis of obesity in children and could affect the lipid metabolism in Chinese obese children.

Keywords: children;obesity;protein tyrosine phosphatase 1B;single nucleotide polymorphism;leptin;insulin

收稿日期 2009-12-18 修回日期 网络版发布日期

DOI: 10.3969/j.issn.1672-7347.2010.

基金项目:

通讯作者:

作者简介:

作者Email: wujing0731@hotmail.com

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1209-1215