遗传 2004 26 (2): 155-159 ISSN: 0379-4172 CN: 11-5450/R

ZFP161基因与中国高度近视人群的相关性研究 Variations of the Zinc Finger Protein 161 gene in Chinese with or Without High Myopia

王国兵1, 张清炯1, 肖学珊1, 李家璋2, 张丰生3, 黎仕强1, 李 玮4, 李 鸵2, 贾小云1, 郭 莉1, 郭向明1
WANG Guo-Bin1, ZHANG Qing-Jiong1, XIAO Xue-Shan1, LI Jia-Zhang2, ZHANG Feng-Sheng3, LI Shi-Qiang1, LI Wei4, LI Tuo2, JIA Xiao-Yun1, GUO Li1, GUO Xiang-Ming1
1.中山大学中山眼科中心暨卫生部眼科学实验室眼遗传分子生物学室，广州 510060； 2.湖北恩施市航空路90号恩施市人民医院眼科，恩施 445000；3.内蒙古包头市沙河镇朝聚眼科医院，包头 014060；4.深圳市笋岗西路深圳市第二人民医院，深圳 5180351. Ocular Genetics and Molecular Biology, National Ophthalmological Labs and Zhongshan Ophthalmic Center, ZhongShan University, Guangzhou 510060, China；2. Department of Opthalmolgy, The people's Hospital of Enshi Autonomous Prefecture, 90 Hangkong Street, Enshi, Hubei 445000, China；3. Chaoju Eye Hospital, Shahe Town, Baotou, Inner Mongolia 014060, China；4. Shenzhen 2nd People's Hospital, Sungang Street, Shenzhen 518035, China

收稿日期 修回日期 网络版发布日期 接受日期

摘要 为了探讨ZFP161基因与高度近视的相关性，从而寻找高度近视的致病基因，以来自不同地区和家系的中国单纯性高度近视先证者204例和排除高度近视及相关疾病的正常人116例为材料，采用PCR-SSCP法检测病例组及正常人群外周血白细胞基因组DNA中ZFP161基因2个外显子是否存在基因突变，对存在突变的外显子区域经克隆测序后确定变异性质，结合对照组及家系分析确定ZFP161基因突变与高度近视的相关性。结果表明：1.ZFP161基因内含子1第58号碱基前存在AT序列插入突变，即IVS1 58～59突变(1/204)，该突变仅存在于高度近视先证者中；2.ZFP161基因外显子2的第168号碱基由C颠换为A，即Ala56Ala突变(Codon56 GCC→GCA, Ala56Ala)(5/204)，该突变存在于正常人群中（3/116），亦存在于患者的正常亲属中；结合正常对照和家系分析，初步排除ZFP161基因与中国单纯性高度近视之间的相关性。Abstract: To investigate the association between variations of ZFP161 gene and high myopia, A total of 204 probands with simple high myopia(≤-6.0 dipoters) were collected while 116 normal persons from different families without high myopia or related disease were used as controls. Genomic DNA was prepared from the peripheral leucocytes. The coding sequences of ZFP161 gene in 320 subjects were analyzed by using exon-by-exon PCR-heteroduplex-SSCP analysis. Identification of the Variations by cloning and sequencing, combinated with controls and family analysis, was used to disclose the correlation between ZFP161 gene and high myopia. A mutation of ZFP161 gene was identified as an insertion of AT before the 58th nucleotide of intron 1 (IVS1 58-59)(1/204) and a variation of ZFP161 gene was identified as a heterozygous C to A of the 168th nucleotide in exon 2 (Codon56, GCC→GCA, Ala56Ala). Ala56Ala is a non-sense mutation identified in 5 of the 204 patients and 3 of 116 controls. No evidence shows that these variations are responsible for high myopia.

关键词 高度近视 ZFP161基因 PCR-SSCPKey words high myopia ZFP161 PCR-SSCP
分类号

Abstract

Key words

DOI:

通讯作者