

重建邻接关系树评估原发性高血压患者的遗传性 Evaluation of Inheritable Character in Essential Hypertension through Reconstruction of Neighbor-Joining Tree

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摘要

探讨原发性高血压患者的遗传性, 评估线粒体D环控制区基因变异在高血压发病中的作用。提取原发性高血压患者和正常血压人群DNA各20例, 用3对交叉重叠引物扩增全部线粒体控制区D环基因, 直接基因测序并重建邻接关系树, 分析原发性高血压的基因变异特点。结果发现部分高血压病患者具有明显群聚倾向, 与正常血压人群和其他无群聚倾向的高血压患者比较, 存在高频率、高密度的D环控制区基因变化 ($P<0.01$), 尤以np152T→C、np182C→T、np189A→G、np247G→A、np16187C→T、np16189T→C、np16264C→T、np16270C→T和np16311T→C等多态性变化显著, 并因此造成np16184~16193微卫星区域多聚C长度改变。本研究提示部分高血压病患者有群聚现象, 基因型np152C、np182T、np247A、np16187T、np16189C、np16264T、np16270T和np16311C可能是此聚类族高血压患者的易感遗传标记。Abstract: To explore the inheritable character in essential hypertension and to evaluate the role of mitochondrial DNA (mtDNA) variations of the D-loop region in the pathogenesis of hypertension, the entire genome of the D-loop region from the hypertensive and the normotensive (20 cases, each) was amplified using 3 pairs of overlapping primers and followed by sequencing. We reconstructed the Neighbor-Joining tree and analyzed the mtDNA variations in the D-loop region. The results exhibited that one clustering branch harbored some hypertensive, who had significantly higher frequency and density of mtDNA variations (both $P<0.01$), especially the polymorphisms of np152T→C, np182C→T, np189A→G, np247G→A, np16187C→T, np16189T→C, np16264C→T, np16270C→T and np16311T→C. This study suggested that there was an aggregative phenomenon in some hypertensive. The genotypes of np152C, np182T, np247A, np16187T, np16189C, np16264T, np16270T and np16311C may be potential genetic markers for susceptibility to hypertension.

关键词 [线粒体基因](#) [D环区](#) [原发性高血压](#) [遗传](#) Key words [mitochondrial DNA](#) [D-loop region](#) [essential hypertension](#) [genetic](#)

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