

血红蛋白E纯合子生化遗传学研究1)

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摘要 本文报告广西地区发现的一个壮族异常血红蛋白家系的研究结果。电泳分析表明, 这种异常血红蛋白是一种慢速血红蛋白, 其电泳迁移率和HBA2相同。血红蛋白的分子杂交和PCMB解离说明是β链第26位的谷氨酸为赖氨酸所替代(βGlu→Lys), 因此称为HbE。HbE在这个家系中至少已经遗传了二代, 共有10例患者, 其中3例是HbE纯合子, HbE含量达90%以上, 其余7例是HbE杂合子。本文对HbE纯合子的产生以及HbE的地理分布和民族起源问题进行了讨论。

关键词

分类号

A STUDY ON BIOCHEMICAL GENETICS OF THE HEMOGLOBIN E HOMOZYGOTE

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Abstract

This paper presents the results of a study of abnormal hemoglobin on a family of Chuang ancestry in Guangxi region. Electrophoresis analysis reveals that the abnormal hemoglobin is a slow-mowing one. Its electrophoretic mobility is the same as that of HbA. From molecular hybridization and PCMB dissociation of the hemoglobin, the β-chains of this hemoglobin are found to be abnormal. Fingerprinting and amino acid analysis of the hemoglobin indicate that a structural change has occurred and β26 glutamic acid is replaced by lysine (β26 Glu→Lys) known as HbE.

HbE is noted at least for two successive generations with 10 persons affected in this family. Three of them are found to be HbE HOMOZYGOTES, with HbE quantity being higher than 90%. The rest are of HbE heterozygotes. The occurrence of HbE homozygote and the relation between geographic distribution of HbE and the origin of Race Chuang are discussed.

Key words

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