中国人血红蛋白New York的研究

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摘要 本文报告在中国大陆发现的10个快速异常血红蛋白系的研究结果,这种异常血红蛋白的电泳迁移移率属于 K型,血红蛋白稳定性轻度降低,血红蛋白变型的结构分析,包括珠蛋白肽链的分离,氨基乙基化β链的酶解和指 纹分析,以及异常肽段的氨基酸组成和顺序测定,证实其β链113位(G15)的缬氨酸被谷氨酸替代,称为血红蛋白 New York (αβ(G15) ValξGlu). 在10个家系的调查中共发现35例Hb New York患者,他们都没有临床症状。其中2例 是Hb New York和β°地中海贫血基因的双重杂合子,血红蛋白New York的含量达90%以上;其余患者都是Hn New York杂合子,血红蛋白New York 含量大约50%。从血红蛋白New York的地理分布可以看出,这种异常血红蛋白在我国南方发生频率较高,接近0.4%。

关键词

分类号

A Study on Hemoglobin New York in China

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Abstract

This paper presents the results of a study of 10 families with a fast abnormal hemoglobin found in China Mainland. The abnormal hemoglobin, with slightly reduced stability, was identified as the K type hemoglobin according to its electrophoretic mobility. From the structure analyses of the variant, including globin chain separation, tryptic digestion of the aminoethylated $|\hat{\mathbf{A}}|$ chain, fingerprinting of the tryptic digest, as well as amino acid composition and sequence analysis of the abnormal peptide, it was indicated that its $|\hat{\mathbf{A}}|$ 113 £ÛG15 £Ý valine was replaced by glutamic acid, and hence known as hemoglobin New York .

In the 10 families studied,35 individuals were found to be the Hb New York s ubjects, all healthy in appearance. Among them two subjects, with hemoglobin New Yo

rk over 90%,were the double heterozygote for hemoglobin New York and ${}^{{}_{|}}\!\hat{A}_{\hat{I}}\tilde{a}$ thalas

semia genes; the remainders were the heterozygote for the Hb New York gene only a

nd had the abnormal hemoglobin about 50%. From the geographical distribution of H b New York, it is clear that Hb New York occurs mostly in the southern parts of C hina, with a frequency of about 0.4%.

Key words

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