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论著

再生障碍性贫血患者PIG-A基因外显子2、4、5突变及粒细胞CD55、CD59的表达

李玉云1，昝丽娜2，王卫国3

1.蚌埠医学院临床检验教研室; 2. 蚌埠医学院临床检验诊断实验中心,安徽 蚌埠 233030;3.阜阳市人民医院检验科,安徽 阜阳 236003

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摘要 【摘要】背景与目的: 探讨再生障碍性贫血(aplastic anemia,AA)患者PIG-A基因外显子2、4、5的突变及粒细胞表面CD55、CD59锚蛋白表达的情况。 材料与方法: 从30例AA患者及20例正常对照组人群外周血提取基因组DNA,采用PCR扩增PIG-A基因外显子2、4、5,再将纯化的PCR产物双向测序检测基因序列;并用流式细胞术检测两组人群外周血粒细胞中CD55、CD59的表达。 结果: 30例AA患者中11例发现PIG-A基因外显子2突变,包括碱基替代、缺失、插入;PIG-A基因外显子4和外显子5突变各5例,仅为碱基替代;共5例患者有2个或2个以上外显子存在突变,正常对照组未发现突变。粒细胞CD55和CD59的表达率在AA患者PIG-A基因突变者中分别为(86.57±5.90)%、(88.17±5.90)%,在PIG-A基因未突变者中分别为(91.87±4.79)%、(94.24±3.76)%,均较正常对照组(97.86±1.52)%、(98.82±1.42)%显著降低(P均<0.05)。 结论: 再生障碍性贫血患者存在PIG-A基因突变和粒细胞CD55、CD59表达缺失的现象,提示再障患者可能存在造血的克隆源性异常。

关键词 【关键词】再生障碍性贫血 PIG-A 外显子 CD55 CD59

Mutations of PIG-A gene Exons 2, 4, 5 and Granulocytic Expressions of CD55 and CD59 in Patients with Aplastic Anemia

LI Yu-yun1,ZAN Li-na2,WANG Wei-guo3

1. Department of Clinical Examination, Bengbu Medical College; 2 .Research Center of Clinical Laboratory Science, Bengbu Medical College, Bengbu 233030;3. Department of Examination,People's Hospital of Fuyang,Fuyang 236003, Anhui,China

Abstract 【ABSTRACT】BACKGROUND AND AIM: To explore mutations of PIG-A gene exon2, exon4, exon5 and expression of CD55 and CD59 in granulocytes of patients with aplastic anemia. MATERIALS AND METHODS: Genomic DNA from peripheral blood of 30 aplastic anemia patients and 20 normal controls were extracted, and PIG-A gene exon2, exon4, exon5 were then examined with polymerase chain reaction(PCR), nucleotide sequences were analyzed by bidirectional sequencing after PCR products were purified. The expressions of CD55 and CD59 in granulocytes from peripheral blood of the two groups above were detected by flow cytometry. RESULTS: The mutations of PIG-A gene exon2 including base substitution, deletion, insertion occurred in 11 of 30 aplastic anemia patients , the mutations of exon4, exon5 were also found in five aplastic anemia patients, with only base substitution. Two or more exon mutations were found in 5 aplastic anemia patients, but the normal controls had no in mutations. The expression of CD55 and CD59 were (86.57±5.90)% and (88.17±5.90)%,respectively, in aplastic anemia patients with PIG-A gene mutation;and (91.87±4.79)% and (94.24±3.76)%, respectively, in aplastic anemia patients with no PIG-A gene mutation. Both were significantly decreased(P<0.05)compared with (97.86±1.52)% and (98.82±1.42)%, respectively, in granulocytes of normal control group. CONCLUSION: The PIG-A gene mutations and decreased expressions of CD55 and CD59 in granulocytes may suggest clonal hematopoietic in patients with aplastic anemia.

Keywords 【KEY WORDS】 aplastic anemia PIG-A exon CD55 CD59

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通讯作者 李玉云 bbmcliyuyun@163.com