

研究论文

## 鉴定9个新的RHD基因mRNA可变剪接体

许先国, 吴俊杰, 洪小珍, 朱发明, 严力行

浙江省血液中心输血研究所, 卫生部血液安全研究重点实验室, 杭州 310006

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**摘要** 为了研究各种RHD基因mRNA可变剪接体的基因结构, 应用逆转录聚合酶链反应(RT-PCR)检测正常人脐血样本RHD mRNA, 对RHD cDNA进行TA克隆和序列分析, 对各可变剪接体的剪接位点进行DNA序列分析, 并将RHD mRNA进行表达序列标签(ESTs)分析。结果在28个阳性克隆中, 除全长RHD cDNA外, 共检测到12种(包括9种新的)RHD可变剪接体, 发现外显子遗漏、5'和3'剪接位点变异3种剪接形式, 涉及外显子2~9, 其中6种新的剪接体同时存在RHD和RHCE基因同源杂交现象。ESTs分析还检索到内含子保留形式的剪接体。研究表明, RHD基因mRNA存在复杂的可变剪接机制, 除已报道的剪接体外, 检测到9种新的RHD可变剪接体, 并发现了可变剪接和同源杂交并存现象。

**关键词** [RHD基因](#) [可变剪接/选择性剪接](#) [Rh血型](#)

**分类号** [R457.1+1](#)

## Identification of Nine Novel Alternative Splicing Isoforms of RHD mRNA

XU Xian-Guo, WU Jun-Jie, HONG Xiao-Zhen, ZHU Fa-Ming, YAN Li-Xing

Institute of Transfusion Medicine, Blood Center of Zhejiang Province, Key Laboratory of Blood Safety Research, Ministry of Health, Hangzhou 310006, China

### Abstract

**Abstract:** To investigate the gene structures of RHD mRNA alternative splicing isoforms, total RNA was extracted from normal cord blood samples and RHD mRNA were detected using reverse transcription and polymerase chain reaction (RT-PCR) method. The PCR products were cloned and directly sequenced. The splicing signals of various isoforms were also analyzed by sequencing of exon-intron boundaries using RHD gene specific primers, and the expressed sequence tags (ESTs) databank was screened to find out other RHD alternative splicing isoforms. Nine novel and three recurrent RHD alternative splicing isoforms were identified, among which six isoforms were RHD-RHCE hybrid genes. Three types of alternative splicing were found including exon skipping, alternative 5' splice sites and alternative 3' splice sites. Exons 2 to 9 were involved in RHD mRNA alternative splicing. Two other isoforms of intron retention type were also found in human ESTs databank. Thus, the mRNA splicing mechanism of RHD gene was very complex, and nine novel alternative splicing isoforms were identified including several isoforms based on homologous RHD-RHCE hybrid gene.

**Key words** [RHD gene](#) [alternative splicing](#) [Rhesus blood group](#)

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