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BRIEF REPORT — POLYMORPHISM REPORT

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A new single-nucleotide polymorphism in the seventh component of complement (*C7*) gene

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Abstract A new single-nucleotide polymorphism has been found in the 3′ untranslated region of the complement component *C7* gene. It is present with similar frequencies in the Japanese and Germans. This polymorphism would be a useful marker in the genetic study of C6 and C7 deficiencies.

Key words Complement · C7 · Mutation · Polymorphism · Population genetics

Introduction

The seventh component of complement (C7) is a precursor protein involved in the formation of the membrane attack complex for the lysis of cells. C7 deficiency is associated with recurrent systemic infections (Tedesco et al. 1993). The *C7* gene is located close to the *C6* gene in a tail-to-tail arrangement, and they are very similar in organization to each other (Hobart et al. 1995). We report here a new polymorphism in the *C7* gene due to an A-to-C transversion at nucleotide 2546 of the cDNA sequence published by DiScipio et al. (1988), i.e., at the 14th nucleotide downstream from the stop codon.

Primers for the polymerase-chain reaction (PCR) of exon 17

The primers published by Nishizaka et al. (1996) were used:
C7-17F: 5′-CTCCACAATGTACCATTAAGC-3′
C7-17R: 5′-TGTGCAGATGTTTTCACTCAG-3′

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Polymorphism and allele frequency

NcoI polymorphism. *Nco*I digestion produced 234-bp and 59-bp fragments in the 2546A allele having the *Nco*I site, while the digestion detected a 293-bp fragment in the 2546C allele that lacks the recognition site.

Allele frequencies. The allele frequencies were determined by polymerase chain reaction-single-strand conformation polymorphism (PCR-SSCP) analysis of genomic DNA from 60 unrelated Japanese and 60 unrelated German individuals (Table 1). No difference in the distribution of allele frequencies was observed between them. The polymorphism information content (PIC) was 0.36.

Chromosome localization. The human *C7* gene has been assigned to chromosome 5p12–14 (Jeremiah et al. 1990).

Mendelian inheritance. Codominant segregation was shown in four two-generation families.

Other comments. PCR was performed in a volume of 50 μl containing 100 ng genomic DNA, 10 mM Tris-HCl (pH8.3), 50 mM KCl, 2 mM $MgCl_2$, 200 μM dNTPs, 10 pmol of each primer, and 1 unit of *Taq* polymerase. Cycle conditions were 94°C for 5 min, then 30 cycles of 94°C for 1 min, 55°C for 1 min, 72°C for 1 min, with a final extension step of 5 min at 72°C, in a Programmable Thermal Controller (MJ Research, Watertown, MA, USA). The PCR products (293 bp) were digested with *Nco*I and subjected to electrophoresis on a 3% Agarose 21 gel (Nippon Gene, Toyama, Japan). SSCP analysis was carried out by subjecting denatured PCR products to electrophoresis on polyacrylamide gels (12%T, 3%C, 1×Tris-borate-ethylenediaminetetraacetic acid [EDTA] [TBE] buffer) at 200 V for 3 h at 4°C, using 0.5× TBE as a tank buffer. Then band patterns were visualized by silver stain. The polymorphism in the *C7* gene may be of interest in the study of human C6 and C7 deficiencies.

Table 1. Polymorphism at nucleotide position 2546 of the complement *C7* gene and gene frequencies in Japanese and German populations

		Genotypes			Gene frequencies	
Population	*n*	A/A	A/C	C/C	2546A	2546C
Japanese	60	9	29	22	0.39	0.61
German	60	7	33	20	0.39	0.61

Heterozygosity, 0.48; polymorphism information content, 0.36

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Erratum

□ In the original article "Position-independent human β-globin gene expression mediated by a recombinant adeno-associated virus vector carrying the chicken β-globin insulator" by T. Inoue et al. [J. Hum Genet. (1999) 44:3, 152–162], Fig. 4A, B (p. 159) , Fig. 6 (p. 160) and figure legend of Fig.6 were reproduced incompletely. The publisher apologizes to the authors. They should have been printed as follows:

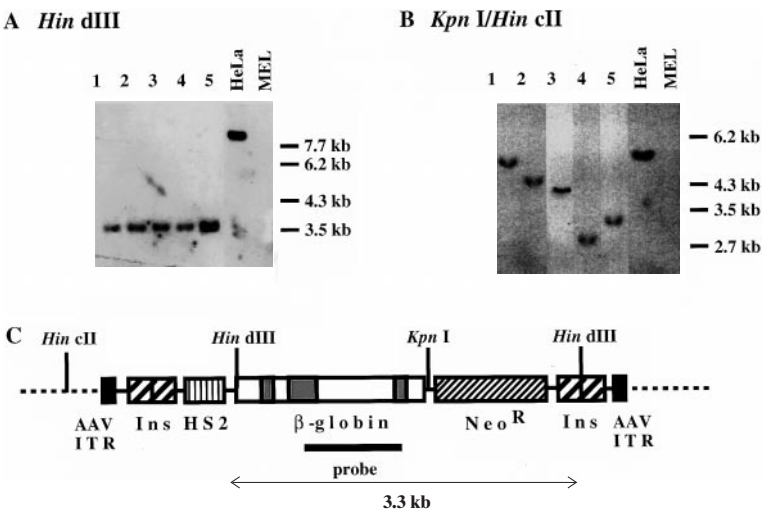

Fig. 4A-C. Southern blot analysis of **A** *Hin* dIII and **B** *Kpn* I and *Hin* cII -digested DNAs from rIns/HS2/Ins clones. DNAs extracted from five independent neomycin-resistant clones of rIns/HS2/Ins were submitted to Southern blot analysis. *Lane HeLa* contains *Hin* dIII- or *Kpn* I and Hin cII-digested DNA from HeLa cells, as a positive control and lane MEL contains *Hin* dIII- or *Kpn* I and *Hin* cII-digested DNA from MEL cells, as a negative control. The human β-globin probe was prepared by radiolabelling a 917-bp fragment flanked by *Bam* HI and *Eco* RI restriction sites. *Tick marks* indicate position of the Lambda *Sty* I marker. **C** Schematic of the integrated rIns/HS2/Ins genome. *Hin* dIII digestion generates a 3.3-kb insert containing the human β-globin gene. Double digestion with *Kpn* I and *Hin* cII results in a junction fragment hybridized with the human β-globin probe

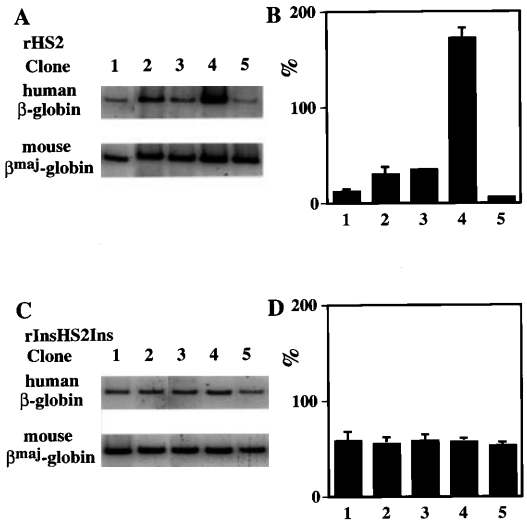

Fig. 6. Expression of the human β-globin gene transduced by rAAV, with and without the insulator in MEL clones. RT-PCR products of RNA extracted from five individual clones with a single unrearranged rAAV vector genome for each construct are shown, A, B rHS2 clones; C, D rIns/HS2/Ins clones. The relative amount of human β-globin mRNA is expressed as a percentage ratio of human β-globin RT-PCR products to mouse bmaj-globin RT-PCR products. The assay was done in triplicate on individual clones