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Localization of a Human-Specific Retroposon (SINE-R.C2) to Chromosome 6p21.31 by Radiation Hybrid Mapping

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Abstract A human-specific retroposon SINE-R.C2 has been derived from a human endogenous retrovirus HERV-K10. It is absent in the genome of nonhuman primates and present within the third intron of the human C2 gene that is located in the class III region of the major histocompatibility complex. In the present study, we determined the regional location of the human C2 gene. The analysis of the Genebridge 4 radiation hybrid mapping panel using PCR amplification located the C2 gene between D6S1422 (10.1 cR) and CHLC.GATA4A03 (21.3) with a lod score of > 3.0. This allowed us to localize C2 gene on the human chromosome 6 band p21.31.

Key words: Human chromosome, Gene mapping, Retroposon, SINE-R.C2

Introduction

A human-specific retroposon SINE-R.C2 has been mapped to intron 3 of the C2 gene [1,2]. The second component of complement, C2 gene, is located in the class III of the major histocompatibility complex on the short arm of human chromosome 6 [3]. The C2 gene is composed of 18 exons and spans about 18 kb of DNA [4]. The SINE-R.C2 is a member of a large SINE-type retroposon family. These retroposons derive from the 3′ long terminal repeat and the small upstream regions of the human endogenous retrovirus HERV-K10 and are present at 4,000 to 5,000 copies per haploid human genome [5]. The HERV-K10 found to be 9.5 kb, present at 50 copies per haploid genome, and homologous to mouse mammary tumor virus (MMTV) [6]. The HERV-K10 was present in all primates except for New World monkeys, whereas SINE-R.C2 is present only in human. SINE-R-type retroposons were present in the human, chimpanzee, gorilla, orangutan, and gibbon genome by Southern blot and PCR analyses [2,7]. Here we report the precise localization of the C2 gene to the human chromosome 6 band p21.31 based on PCR analysis of the Genebridge 4 radiation hybrid mapping panel.

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Materials and Methods

A Genebridge 4 radiation hybrid mapping panel was obtained from the MRC Human Genome Mapping Project Resource Centre (HGMP-RC, Cambridge). The panel samples were subjected to PCR amplification. The PCR products were analysed by 2.5% agarose gel electrophoresis and ethidium bromide staining.

The cDNA sequence of the C2 gene was obtained from the GenBank under accession number L09706 (12293 bp). A 212-bp fragment of the C2 gene was amplified using primers HS1018 (5′-CTCGAGAGACTTGCAGCTTAT-3′, bases 1-21) and DS1019 (5′-ACCCACCCAATATGTAG-GAAAT-3′, bases 191-212). The PCR conditions followed were those of Kim et al. [8] with an annealing temperature of 57℃.

Results and Discussion

The second component of complement, the human C2 gene, is located in the class III of the major histocompatibility complex on the short arm of chromosome 6 [3]. A human-specific retroposon SINE-R.C2 has been mapped to intron 3 of the C2 gene [1,2]. To determine the precise localization of the human C2 gene, the radiation hybrid panel was analysed by a PCR approach. PCR amplification using DNA derived from a Genebridge 4 radiation hybrid mapping panel with the specific primers, HS1018 and DS1019, designed from the 5′ untranslated region of human C2 gene showed the expected product of 212 bp. Eighteen out of the 93 hybrids were positive, 66 were negative, and in 9 the results were not clear. Data vectors were transmitted to the Whitehead Institute/MIT Radiation Hybrid Mapper soft-

ware (http://carbon.wi.mit.edu:8000/cgi-bin/contig/rhmapper.pl). The result indicates that the human C2 gene is on the short arm of chromosome 6 between D6S1422 (10.1 cR) and CHLC.GATA4A03 (21.3) with a lod score of > 3.0. This region is located in human chromosome 6 band p21.31 according to the GDB Internet site map (http://www.gdb.org/jmqp/quereByPosn.html).

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