1992

A new taql allele detected by the CRI-R227 (D4S101) probe in Pima Indians

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Abstract

[No abstract available]

Keywords

taql, allele, detected, cri, r227, d4s101, probe, pima, indians

Disciplines

Medicine and Health Sciences

Publication Details

Choi, M., Prochazka, M., Thuillez, P. Lillioja, S. (1992). A new taql allele detected by the CRI-R227 (D4S101) probe in Pima Indians. Nucleic Acids Research, 20 (5), 1157-1157.

PCR detection of an insertion/deletion polymorphism in intron 1 of the HRAS1 locus

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Intron 1 of the c-Ha-Ras-1 gene (HRAS; MIM N 190020) contains a tandemly repeated hexanucleotide sequence — based on the consensus GGGCCT — which show length polymorphism after digestion with the restriction enzyme PstI (1). Here we describe a protocol for direct detection of alleles from this region by PCR. EMBL accession no. K00654, bp 244–361.

PCR Primers:
Sense oligo: 5'CTGTGGGTTTGCCCTTCAGA
Antisense oligo: 5'CTCCTACAGGGTCTCCTGCC

Polymorphism: Three different alleles, can be identified after DNA amplification. Two alleles had been previously described: allele F1, containing 4 repeats, and allele F2, containing 2 repeats (1), which correspond to 118 and 106 bp amplification products, respectively. A third allele — F3 — was identified in the present survey. This is a 112 bp fragment, corresponding to 3 repeat units.

Allele Frequencies: F1 = 0.61; F2 = 0.28; F3 = 0.11, based on analysis of 146 unrelated Italian individuals. Observed heterozygosity = 0.52.

Mendelian Inheritance: Codominant segregation demonstrated in 5 two-generation families (32 individuals).

Chromosomal Localization: 11p15.5 (2).

PCR Conditions: Standard reaction mixtures contained 0.5 μg of genomic DNA, 0.25 μM each primer, 0.2 mM each dNTP, 1.5 mM $MgCl_2$, 50 mM KCl, and 10 mM Tris at pH 8.3. Thirty amplification rounds were performed under the following conditions: 1 minute denaturation at 95°C, 1 minute annealing at 58°C, and 3 minute extension at 70°C. Alleles were visualized by ethidium bromide staining. An extra band of lower mobility was often visualized in heterozygous individuals. This phenomenon was attributed to heteroduplex formation, as confirmed by denaturation/renaturation experiments.

Comments: The relatively high heterozygote frequency makes this system a first choice for studies of genetic linkage and of reduction to homozygosity in tumor cells at the HRAS locus.

Acknowledgements: This work was supported by the A.I.R.C., Milano.

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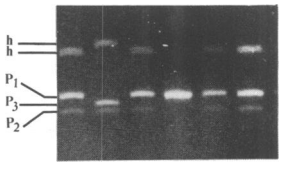

A new TaqI allele detected by the CRI-R227 (D4S101) probe in Pima Indians

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Source and Description: Probe CRI-R227 is a 15–20 kb fragment isolated from a Charon 4A phage library of human genomic DNA (1). A 3.7 kb fragment of EcoRI digestion of CRI-R227 was used because it was as informative as the whole probe.

Polymorphism: CRI-R227 was originally reported to detect one 2-allele polymorphism by EcoRI (A1: 5.1 kb, A2: 3.7 kb) and two 2-allele polymorphisms by TaqI (B1: 5.3 kb, B2: 5.0 kb, C1: 3.3 kb, C2: 2.4 kb) in 5 unrelated Caucasians (2). In Pima Indians (n = 161) a new and common TaqI allele of 5.1 kb size fragment (C3) has been detected.

Frequency: Allele frequencies were determined from 63 unrelated Pima Indians. Haplotype frequencies were determined for 38 of these subjects in whom the haplotypes could be assigned unambiguously. Among an additional 98 first degree relatives of the above 63 subjects, there are no unambiguous B2C2 or B2C3 haplotype combinations.

Allele System	Size (kb)	Freq.	Haplotypes	Obs. Freq.	Exp. Freq.
B	5.3 (B1)	.47	B1 C1	.05	.26
	5.0 (B2)	.53	B1 C2	.13	.08
C	3.3 (C1)	.56	B1 C3	.26	.13
	2.4 (C2)	.17	B2 C1	.55	.30
	5.1 (C3)	.27	B2 C2	.00	.09
			B2 C3	.00	.14

Mendelian Inheritance: Co-dominant segregation of the C alleles was observed in a three generation family (figure).

Chromosomal Location: Probe CRI-R227 (D4S101) maps to chromosome 4q (3).

Probe Availability: Probe CRI-R227 was purchased from Collaborative Research Inc.

Other Comments: A1, A2 alleles of EcoRI polymorphism were completely associated with B2, B1 alleles of TaqI polymorphisms, respectively, making typing for A1, A2 superfluous.

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