

Dinucleotide repeat polymorphism in the human interleukin 1, alpha gene (IL1A)

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Source/Description: An (AC)_n repeat sequence is located at position 7,695 of the human interleukin 1 alpha (IL1A) gene on chromosome 2q12-q21 (1). PCR primers have been designed (IL1AAC1 5'GCCTAGTGAGTGTGGAAGACATTG3', IL1AAC2 5'CAGCACTGGTTGGTCTTCATCTTG3') to amplify a 334 bp region containing the polymorphic repeat.

Frequency: Twenty unrelated individuals demonstrated six alleles. The observed heterozygosity was 74%.

Allele	Frequency
D1	0.44
D2	0.21
D3	0.03
D4	0.08
D5	0.16
D6	0.08

Chromosomal Localization: The human interleukin 1 alpha (IL1A) gene has been localized to chromosome 2q12-q21 (2, 3).

Mendelian Inheritance: Co-dominant segregation was observed in three three-generation families.

Other Comments: The PCR reactions were performed on 200 ng genomic DNA with 10 ng ³²P-labeled IL1AAC1 primer and 10 ng unlabeled IL1AAC2 primer. These reactions were carried out as described previously (4). The amplified products were run on a 5% polyacrylamide, 32% formamide sequencing gel at 80 watts constant power for approximately 4 hours in 1×TBE buffer.

References: 1) Furutani, Y. *et al.* (1986) *Nucl. Acids Res.* **14**, 3167–3179. 2) Modi, W.S. *et al.* (1988) *Genomics* **2**, 310–314. 3) Lafage, M. *et al.* (1989) *Blood* **73**, 104–107. 4) Theune, S. *et al.* (1991) *Genomics* **9**, 511–516.

Dinucleotide repeat polymorphism in the human surfactant-associated protein 3 gene (SFTP3)

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Source/Description: Two (AC)_n repeat sequences are located at positions 2,988 and 3,050 of the human surfactant-associated protein 3 gene on chromosome 2 (1). PCR primers have been designed (SFTP3AC1 5'CAGCACCTATCCAGACACATACA3', SFTP3AC2 5'TGTGTGTGAGAGTGAGGGTGTAAG3') to amplify a 321 bp region containing the polymorphic repeats.

Frequency: Twenty unrelated individuals demonstrated five alleles. The observed heterozygosity was 50%.

Allele	Frequency
B1	0.36
B2	0.30
B3	0.28
B4	0.03
B5	0.03

Chromosomal Localization: The human surfactant-associated protein 3 (SFTP3) gene has been localized to chromosome 2 (2).

Mendelian Inheritance: Co-dominant segregation was observed in three three-generation families.

Other Comments: The PCR reactions were performed on 200 ng genomic DNA with 10 ng ³²P-labeled SFTP3AC1 primer and 10 ng unlabeled SFTP3AC2 primer. These reactions were carried out as described previously (3). The amplified products were run on a 5% polyacrylamide, 32% formamide sequencing gel at 80 watts constant power for approximately 4 hours in 1×TBE buffer.

References: 1) Pilot-Matias, T.J. *et al.* (1989) *DNA* **8**, 75–86. 2) Emrie, P.A. *et al.* (1988) *Somat. Cell Mol. Genet.* **14**, 105–110. 3) Theune, S. *et al.* (1991) *Genomics* **9**, 511–516.