DNA amplification for detection of the XV-2c polymorphism linked to cystic fibrosis

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SOURCE/DESCRIPTION: A probe designated XV-2c was previously reported to detect a polymorphism which shows linkage disequilibrium with the cystic fibrosis (CF) locus [1]. We have identified oligonucleotides to analyze the RFLP by DNA amplification.

POLYMORPHISM: The alleles are as for the RFLP reported by Southern blot [1]. The amplified product is 1.0 kb in size and is cut into fragments of 0.6 and 0.4 kb by *TaqI*. Allele 1 = absence of cut site, allele 2 = presence of cut site.

FREQUENCY: North American normal chromosomes: A1 = 0.44, A2 = 0.56.
North American CF chromosomes: A1 = 0.93, A2 = 0.07

NOT POLYMORPHIC FOR: Not tested.

CHROMOSOMAL LOCALIZATION: Tightly linked to CF locus on 7q [1].

MENDELIAN INHERITANCE: Codominant inheritance in over 20 families with tight linkage to cystic fibrosis locus.

PROBE AVAILABILITY: Not needed for amplification. Contact R. Williamson for original probe [1].

OTHER COMMENTS: Oligonucleotide sequences for amplification are as follows: 5'-GTTGAAGTGAATTGAATG-3' and 5'-GTTCAAACTATGTCAAAG-3'. DNA was amplified with *Taq* polymerase as recommended by the supplier, Cetus Corporation. The figure demonstrates the RFLP after amplification, enzyme digestion and direct visualization with ethidium bromide. M = markers, U = uncut with *TaqI*, C = cut with *TaqI*, and B = blank with no genomic DNA. Genotypes are indicated as (+) for presence of the *TaqI* site and (-) for absence.

REFERENCE: (1) Estivill et al., *Nature* 326:840, 1987.