Additional polymorphism for D7S8 linked to cystic fibrosis including detection by DNA amplificationH.Northrup¹, C.Rosenbloom¹, W.E.O'Brien^{1,2} and A.L.Beaudet^{1,2}¹Institute for Molecular Genetics, Baylor College of Medicine and ²Howard Hughes Medical Institute, Houston, TX 77030, USA

SOURCE AND DESCRIPTION OF CLONE: A cosmid was cloned from a human genomic library using the pJ3.11 probe [1]. A subclone designated pJG3C is a 2.5 kb fragment inserted in the *EcoRI* site of pUC18.

POLYMORPHISM: *PstI* identifies a two allele system with allele 1 (10.5 kb) and allele 2 (6.2 kb and 4.3 kb). The probe also detects the known *MspI* RFLP [1].

FREQUENCY: Estimated from 41 unrelated Caucasians. Strong linkage disequilibrium exists with other polymorphisms at D7S8 such that allele 1 was found with the N haplotype and allele 2 with the O or P haplotype in 71 of 72 chromosomes [2].

Allele 1 (10.5 kb): 0.38

Allele 2 (6.2 and 4.3 kb): 0.62

NOT POLYMORPHIC FOR: 39 other enzymes.

CHROMOSOMAL LOCALIZATION: Within 30 kb of pJ3.11 at D7S8 on chromosome 7q. Linkage data and pulsed field gel mapping of the region are available [3].

MENDELIAN INHERITANCE: Codominant segregation in 26 cystic fibrosis families with tight linkage to CF.

PROBE AVAILABILITY: Contact A.L.B.

OTHER COMMENTS: Sequencing was used to establish conditions for detection by the polymerase chain reaction (PCR). Oligonucleotide sequences for amplification were 5'-CCACTGATACTGTGAGAC-3' and 5'-GTTGTTTCAAGTCACTGC-3'. The figure demonstrates the RFLP after PCR, enzyme digestion and direct visualization with ethidium bromide. M = markers, U = uncut with *PstI*, C = cut with *PstI*, and B = blank with no genomic DNA. Genotypes are indicated as (+) for presence of the *PstI* site and (-) for absence.

REFERENCES: (1) Bartels et al. *Am J Hum Genet* 38:280, 1986. (2) Spence et al. *Am J Hum Genet* 39:729, 1986. (3) Poustka et al. *Genomics* 2:337, 1988.