Isolation and mapping of a polymorphic DNA sequence pH30 on chromosome 4 [HGM provisional no. D4S139]E.C.B.Milner, C.L.Lotshaw, K.Willems van Dijk, P.Charmley¹, P.Concannon and H.W.Schroeder Jr²

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SOURCE/DESCRIPTION: This clone is a 4.5 kb Sau3A fragment in pUC18 subcloned from a EMBL4 clone containing a 20 kb insert in the EcoRI site identified with the oligonucleotide, H30 (5'-GCCGTGTCCTCGGCTCTCAGG-3').

POLYMORPHISM: TaqI optimally resolves a >20 allele VNTR polymorphism with bands between 4.0 and 25.0 kb. EcoRI, HindIII, BamHI, BglII, PstI, and XbaI also detect the same polymorphism.

HETEROZYGOSITY: With TaqI, 89% heterozygosity was observed in 85 unrelated Caucasians.

CHROMOSOMAL LOCALIZATION: pH30 has been assigned to distal chromosome 4q by multipoint linkage analysis (1). The maximum lod score (3.01, $\theta = 0.162$) was obtained with the locus CRI-L231 (2).

MENDELIAN INHERITANCE: Co-dominant segregation of the VNTR polymorphism has been observed in 18 three generation families and 6 two generation families.

PROBE AVAILABILITY: Contact E.C.B.M.

OTHER COMMENTS: RFLPs observed under normal stringency.

REFERENCES:

1. G.M. Lathrop et al., Am. J. Hum. Genet. 37:482-498 (1985)
2. H. Donis-Keller et al., Cell 51:319-337 (1987)

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