A PstI RFLP for the human retinoic acid receptor in 17q21

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SOURCE/DESCRIPTION: A 1.6 kb KpnI-EcoRI fragment from the cDNA clone p63 (1).

POLYMORPHISM: PstI identifies a two allele polymorphism with bands at 3.0 kb (P1) or 2.6 kb + 0.4 kb (P2). Under medium stringency conditions (see below), invariant bands are detected at 5.7 kb, 3.8 kb, 1.5 kb, 1.4 kb, 1.3 kb, 1.1 kb, 0.9 kb, 0.8 kb and 0.3 kb.

FREQUENCY: Estimated in 26 unrelated caucasians.

P1 (3.0 kb) : 0.19

P2 (2.6 kb + 0.4 kb) : 0.81

NOT POLYMORPHIC FOR: ApaI, ApaLI, BamHI, BanI, BanII, BglIII, BstNI, BstXI, DraI, EcoO109, EcoRI, HindIII, MspI, NheI, PvuII, ScaI, StuI, TaqI, TthIII, XmnI in a minimum of 10 independent chromosomes tested.

CHROMOSOMAL LOCALISATION: The human retinoic acid receptor (hRAR) gene has been mapped in 17q21 by in situ hybridization (2).

MENDELIAN INHERITANCE: Codominant segregation was demonstrated in a large three generation family (28 individuals).

PROBE AVAILABILITY: Available for linkage studies from P. Chambon.

OTHER COMMENTS: Hybridization was performed in the presence of 40 % formamide at 42°C. Medium stringency conditions of washing are defined as 0.5 x SSC - 0.1 % SDS at 60°C. At higher stringency (0.2 x SSC - 0.1 % SDS at 70°C) some of the invariant bands are lost (bands at 1.3 kb and 1.1 kb), which presumably result from cross-hybridization with other member(s) of the nuclear receptor multigene family to which the hRAR belongs.

REFERENCES:

1. Petkovich M., Brand N.J., Krust A., and Chambon P. (1987), *Nature* **330**, 444-450.
2. Mattei M.G., Petkovich M., Mattei J.F., Brand D., and Chambon P. (1988), *Human Genetics*, submitted.