
ApaI dimorphism at the human vitamin D receptor gene locus

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SOURCE/DESCRIPTION pH13, a 2.1kb full length vitamin D receptor cDNA. This probe was isolated from a human breast cancer cell line (T47D) cDNA library, and cloned as an EcoRI fragment into pGEM (1).

POLYMORPHISM ApaI (GGGCCC) (International Biotechnologies, Inc., New Haven, CT) detects four invariant bands at 12.0, 6.0, 3.8, and 1.5kb, and a single two-allele polymorphism with bands at either 7.9kb or 3.0kb.

FREQUENCY Studied in a random population of 153 North Americans

7.9kb allele: $p=0.56\pm0.03$

3.0kb allele: $q=0.44\pm0.03$

CHROMOSOMAL LOCALIZATION Human vitamin D receptor gene localized to chromosome 12 by Southern blot analysis of Human-Chinese hamster cell hybrids (2).

MENDELIAN INHERITANCE Co-dominant inheritance demonstrated in 1 family of 7 individuals.

PROBE AVAILABILITY Requests for probe to J.S. at the above address

REFERENCES 1- Baker A, Shine J, et al. Proc. Natl. Acad. Sci. USA (in press)
2- Shows TB, and Shine J (in preparation)

