An EcoRV RFLP detected by probe pX59 [DXS77]

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SOURCE/DESCRIPTION: An anonymous 0.9 kb EcoRI fragment subcloned into pBR322 (Willard et al, 1983).

POLYMORPHISMS: EcoRV (GAT'ATC) identifies two alleles with bands at 7.5 and 4.3 kb.

FREQUENCY: 30 unrelated Caucasian women tested.

7.5 kb allele (A1): 0.92

4.2 kb allele (A2): 0.08

NOT POLYMORPHIC FOR: BamHI, BglII, BstnI, EcoRI, HindIII, HinfI, MspI, PstI, RsaI, SacI, TaqI, XbaI, and XhoI.

CHROMOSOMAL LOCALISATION: localized to Xp21.1-11.2 using a panel of der(X) containing human-hamster cell hybrids (Diergaarde et al, 1989).

MENDELIAN INHERITANCE: X-linked inheritance was shown in 3 families.

PROBE AVAILABILITY: Contact B.J. Schmeckpeper.

REFERENCE: - Willard H.E., Schmeckpeper B.J., Smith K.D., Goss S.J. Cytogenet Cell Genet 37:609-610, 1983.
- Diergaarde P.J., Wieringa B., Bleeker-Wagemakers E.M., Sims K.B., Breakefield X.O., H.-H. Ropers Human Genetics, in press, 1989.

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