A TaqI RFLP in Xq26-qter detected by pX301b [DXS311]

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SOURCE/DESCRIPTION: A 3.0 kb EcoRI unique fragment subcloned into the EcoRI site of pTZ19R. The probe pX301b is a fragment of a phage clone, λ X301, isolated from a flow-sorted X-chromosome library (Vector: λ gtWES) (1).

POLYMORPHISM: TaqI digests reveal a two-allele polymorphism with a band(s) at either 5.7 kb or 4.3 kb and 1.4 kb. The probe also detects an invariant band at 1kb.

FREQUENCY: Studied in 13 Caucasians: 10 female, 3 male (23 X-chromosomes):

A1	5.7kb allele	0.74
A2	4.3/1.4 kb allele	0.26

NOT POLYMORPHIC FOR: EcoRI, BglII, PstI, SstI

CHROMOSOMAL LOCALIZATION: The probe was localized to Xq26-qter using a panel of somatic cell hybrids deleted for various parts of the X-chromosome. Multipoint linkage analysis (2) in 10 fragile-X families mapped the probe locus 1cM (Z=3.19) proximal to F9 (3).

MENDELIAN INHERITANCE: X-linked codominant segregation pattern observed in 10 families with the fragile-X syndrome.

PROBE AVAILABILITY: Available now.

REFERENCES:

1. Davies KE, Young BD, Elles RG, Hill ME, Williamson R (1981). Cloning of a representative genomic library of the human X-chromosome after sorting by flow cytometry. *Nature* 293: 374-376.
2. Lathrop GM, Lalouel JM, Julier C, Ott J. (1984). Strategies for multilocus linkage analysis in humans. *Proc Natl Acad Sci USA* 81: 3443-3446.
3. Sood R, Mulligan LM, Poon R, White EN, Holden JJA. Genetic mapping of two new DNA markers in Xq26-q28 relative to the fragile-X syndrome locus (in preparation).

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