Isolation and mapping of a polymorphic DNA sequence, DXS312, to Xq27–Xq28


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SOURCE/DESCRIPTION: The parental phage clone was isolated from a human X-chromosome-hamster library constructed in λ Charon 4 (1). Two unique fragments (a HindIII 1.6 kb fragment and a HindIII/EcoRI 1.1 kb fragment) were subcloned into pUC8 and termed pX135f and pX135g, respectively.

POLYMORPHISM: The probe pX135f detects an AvaII RFLP with A1 = 4.0 kb and A2 = 6.6 kb. The probe pX135g detects a different AvaII RFLP with B1 = 5.8 kb and B2 = 3.4 kb.

FREQUENCY: Results from 100 unrelated chromosomes:
- A1 - 4.0 kb allele 0.76

Results from 111 unrelated chromosomes:
- B1 - 5.8 kb allele 0.70

The two RFLPs are in linkage disequilibrium with haplotype frequencies:
- A1 B1 = 0.70
- A1 B2 = 0.06
- A2 B1 = 0.00
- A2 B2 = 0.24

NOT POLYMORPHIC FOR: BamHI, BglII, BstEII, CfoI, EcoRI, HaeIII, HincII, HindIII, Hinfl, KpnI, MspI, NcoI, PstI, PvuII, SstI, TaqI, XbaI and XmnI.

CHROMOSOMAL LOCALIZATION: The probes were localized to Xq26–Xqter using a panel of somatic cell hybrids deleted for various parts of the X-chromosome. The probe pX135f was localized to Xq27–Xq28 by in situ hybridization. Multipoint linkage analysis in 15 fragile-X families placed the probe locus 13 cM proximal to the fragile-X locus (2).

MENDELIAN INHERITANCE: X-linked codominant segregation pattern observed in 15 families with the fragile-X syndrome.

PROBE AVAILABILITY: Available to collaborators.

REFERENCES:


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