A new Xmnl polymorphism for the DMD probe PERT 87-8

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Source/Description: PERT 87-8 is a 1.3 kb intronic fragment from the DMD gene which has been cloned into the XbaI site of pUC18 (1).

Polymorphism: Xmnl identifies a 2 allele polymorphism A1: 9.2 kb A2: 8.8 kb

Frequency: Estimated from a total of 130 chromosomes from an unrelated sample of British Caucasians.
Xmnl AA1: 0.35 AA2: 0.65

Chromosomal Location: PERT 87-8 is a subclone of DXS164 which has been assigned to Xp21.2 (1).

Mendelian Inheritance: Co-dominant segregation in three 2-generation families.

Probe Availability: Contact L.M. Kunkel.

Other Comments: The polymorphism is observed under standard hybridisation and wash conditions.

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Three RFLPs at the D8S586 locus

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Source/Description: Probe pNCO907 contains a 500-bp PstI fragment of pNCO901 inserted into pUC19 as described previously (1).

Polymorphism: EcoRI detects a two allele polymorphism (A1:4.8 kb; A2:1.3 kb). Spl detects a two allele polymorphism (B1: 22.0 kb; B2: 7.0 kb). TaqI detects a four allele polymorphism (C1:11.0 kb; C2:10.0 kb; C3:8.5 kb; C4:6.7 kb).

Frequency: Studied in unrelated Japanese.
A1:0.50 A2:0.50 (33 individuals)
B1:0.21 B2:0.79 (17 individuals)
C1:0.08 C2:0.42 C3:0.09 C4:0.41 (40 individuals)

Not Polymorphic For: PvuII, Mspl, Rsal, or BamHI with DNA from five unrelated individuals.

Chromosomal Localization: Assigned to chromosome 8 using a panel of human-mouse somatic cell hybrids (1).

Mendelian Inheritance: Codominant, independent, segregation of the allele systems A, B and C was observed in one informative family (Figure).

Probe Availability: Available from M. lizuka.

Other Comments: This probe is co-amplified with MYC in COLO320 DM cells but not in HL60 cells (1). The hybridization condition should be highly stringent.

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