A Rsal RFLP at the DXS467 locus

R.G. Lafreniere and H.F. Willard
Department of Genetics, Stanford University, Stanford, CA 94305, USA

Source/Description: cpX12 is a 1.1 kb EcoRI single-copy genomic fragment cloned into pAT153 from a pool of 100 cosmids isolated from a human/hamster somatic cell hybrid containing only the human X chromosome (1).

Polymorphism: cpX12 reveals a two-allele restriction fragment length polymorphism with bands at 0.7 kb and 1.9 kb (A1) or 2.6 kb (A2) after Southern blot analysis of human genomic DNA digested with Rsal.

Frequency: Estimated from 56 unrelated CEPH parents (97 X chromosomes total).
   A1 (0.7 + 1.9 kb) = 0.22
   A2 (2.6 kb) = 0.78
Of 39 females tested, 14 were heterozygous.


Chromosome Localization: Mapped to Xq12 by Southern blot hybridization to a panel of human/rodent somatic cell hybrids (2, 3).

Mendelian Inheritance: X-linked inheritance demonstrated in 14 informative two- and three generation CEPH families.

Probe Availability: Available from Dr. Bert Bakker, Department of Human Genetics, University of Leiden, Leiden, The Netherlands.

Other Comments: Final filter washes in 0.1 x SSC/0.1% SDS at 65°C.


A BstE II RFLP at the DXS153 locus

R.G. Lafreniere and H.F. Willard
Department of Genetics, Stanford University, Stanford, CA 94305, USA

Source/Description: cX37.1 is a 3.3 kb EcoRI single-copy genomic fragment cloned into pAT153 from a cosmid isolated from a human/hamster somatic cell hybrid containing only the human X chromosome (1).

Polymorphism: cX37.1 reveals a two-allele restriction fragment length polymorphism with bands at 3.2 kb (A1) and 5.0 kb (A2) after Southern blot analysis of human genomic DNA digested with BstE II.

Frequency: Estimated from 57 unrelated CEPH parents (97 X chromosomes total).
   A1 (3.2 kb) = 0.22
   A2 (5.0 kb) = 0.78
Of 40 females tested, 15 were heterozygous.

Not Polymorphic For: BamHI, BglII, Dral, EcoRI, EcoRV, HindIII, MspI, PstI, PvuII, Rsal, SstI, TaqI and XbaI in a screen of six unrelated females.

Chromosome Localization: Mapped to Xq12 by Southern blot hybridization to a panel of human/rodent somatic cell hybrids (2, 3).

Mendelian Inheritance: X-linked inheritance demonstrated in 15 informative two- and three generation CEPH families.

Probe Availability: Available from Dr. Bert Bakker, Department of Human Genetics, University of Leiden, Leiden, The Netherlands.

Other Comments: Rare BglIII variant also found with alleles at 5.3 kb (1 of 77 X chromosomes) and ~17 kb (76 of 77 X chromosomes). Final filter washes in 0.1 x SSC/0.1% SDS at 68°C.