A BstXI polymorphism detected by the factor VIII genomic probe p.482.6 (F8C)

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SOURCE/DESCRIPTION:

A 9.6 kb EcoRI fragment in pUC 18 from intron 22 of the factor VIII gene (1,2).

POLYMORPHISM:

BstXI identifies a three allele polymorphism with bands of 2.4 kb (F1), 1.6 kb (F2), or absence of a band (F3), as well as the polymorphism detected by probe 767 (DXS115) (3) with bands at 6.4 kb (A1) and 4.25 kb (A2). Invariant bands of 6.8 kb, 1.8 kb, 1.4 kb and 0.6 kb are also detected.

FREQUENCIES

Studied in 25 unrelated Canadian individuals (31 X chromosomes)

(F1) - 0.48

(F2) - 0.45

(F3) - 0.07

CHROMOSOMAL LOCALISATION:

Chromosome Xq28 (4)

MENDELIAN INHERITANCE:

X-linked segregation was demonstrated in six families of Canadian origin.

PROBE AVAILABILITY:

Through the American Type Culture Collection.

OTHER COMMENTS:

Use of a prehybridization mix of 0.3 M NaCl, 1% SDS, 10% Dextran Sulphate, 100 ug/ml sheared salmon sperm DNA and 25 ug/ul sheared human DNA at 65°C facilitates visualization of this polymorphism.

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