RFLP detected by an X-linked cDNA encoding erythroid-potentiating activity/tissue inhibitor of metalloproteinase (EPA/TIMP)

Sharon J. Durfy\textsuperscript{1}, Steven C. Clark\textsuperscript{2}, Bryan R. G. Williams\textsuperscript{1,3} and Huntington F. Willard\textsuperscript{1,3}

\textsuperscript{1}Department of Medical Genetics, University of Toronto, Toronto, Ontario M5S 1A8, Canada, \textsuperscript{2}Genetics Institute, 87 Cambridge Park Dr., Cambridge, MA 02140, USA and \textsuperscript{3}Research Institute, The Hospital for Sick Children, Toronto, Ontario M5G 1X8, Canada

SOURCE/DESCRIPTION: A full length (1.1 kb) human EPA cDNA clone inserted in p91023(B) at the EcoRI restriction site (1).

POLYMORPHISM: BgllII identifies a two allele polymorphism with bands of approximately 12 kb (A1) or 9.5 and 2.5 kb (A2).

FREQUENCY: A total of 102 X chromosomes from 54 unrelated individuals was examined.
- A1 (12 kb allele): 0.66
- A2 (9.5 and 2.5 kb allele): 0.34

NOT POLYMORPHIC FOR: HindIII, EcoRI, TaqI, RsaI, PstI, BamHI, PvuII or XbaI with a panel of three females and one male.

CHROMOSOMAL LOCALIZATION: Xp11.1 to Xp11.4 (2,3).

MENDELIAN INHERITANCE: Co-dominant segregation shown in one family (see figure).

PROBE AVAILABILITY: For RFLP studies, contact BRGW or HFW.

OTHER COMMENTS: Low background under usual stringency conditions. Final wash at 0.1 x SSC, 65C.


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