The gelsolin (GSN) cDNA clone, from 9q32-34, identifies BclI and StuI RFLPsD.J.Kwiatkowski, L.Ozelius¹, D.Schuback¹, J.Gusella¹ and X.O.Breakefield^{1,2}

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SOURCE AND DESCRIPTION OF CLONE: M1D is a partial length plasma gelsolin cDNA clone (Kwiatkowski et al., 1988). The PstI - PstI fragment, bases 341 - 861, identifies the BclI RFLP; the NcoI-EcoRI 3' fragment, bases 1458-2100, identifies the StuI RFLP.

POLYMORPHISM: BclI identifies a simple two allele polymorphism with bands at 7.0 kb (A1), and 4.9 and 2.1 kb (A2); invariant bands of sizes 7.8 kb and 4.9 (weak) kb are also seen (Fig. 1A). StuI identifies two independent simple two allele polymorphisms with bands at 8.7 (B1) and 7.9 kb (B2), and 2.8 (C1) and 2.6 (C2) kb (Fig. 1B). An invariant band of size 4.0 kb is also seen.

FREQUENCY: For BclI, we examined

20 North Americans: A1 - 0.90, A2 - 0.10

13 Venezuelans: A1 - 0.81, A2 - 0.19.

For StuI, we examined

40 North Americans: B1 - 0.16, B2 - 0.84; C1 - 0.00, C2 - 1.00

16 Venezuelans: B1 - 0.37, B2 - 0.63; C1 - 0.13, C2 - 0.87

14 American Jews: B1 - 0.36, B2 - 0.64; C1 - 0.00, C2 - 1.00.

NOT POLYMORPHIC FOR: Eco RI, BglII, RsaI, TaqI, MspI, HindIII, EcoRV, BamHI, PvuII, HincII, PstI, SacI, Styl, DraI, XmnI, XbaI, and BglI, with DNA from 5 unrelated donors.

CHROMOSOMAL LOCALIZATION: The gelsolin gene (GSN) has been localized to 9q32-34, centromeric to ABL, by somatic cell hybrid DNA analysis, and in-situ hybridization to a normal chromosomal spread and to spreads containing the Philadelphia chromosome [(9;22)(q34;q11)] (Kwiatkowski et al., 1988).

MENDELIAN INHERITANCE: Co-dominant segregation of the A1 - A2 (Fig. 1A) and B1 - B2 (Fig. 1B) polymorphisms was shown in 12 informative families, 251 individuals; co-dominant segregation of the C1 - C2 (Fig. 1B) polymorphism was shown: in 7 families, 200 individuals.

PROBE AVAILABILITY: contact D. J. Kwiatkowski.

REFERENCE: Kwiatkowski et al., Am J. Hum. Genet. 42:565-572, 1988.

ACKNOWLEDGEMENTS: We thank the NIH (HL01582, NS20012), and the Dystonia Medical Research Foundation.