A KpnI polymorphism for the human insulin-responsive glucose transporter gene (*GLUT4*) on chromosome 17

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SOURCE/DESCRIPTION phJHT-3 encodes part of the major insulin responsive glucose transporter expressed in skeletal muscle, heart and adipose tissue. The 1.7 kb EcoRI-linked cDNA insert (vector-pBR327) includes 145bp of 5'-untranslated region, 1326 bp encoding amino acids 1-442 and 267 bp of an intron following codon 442 (T. Kayans et al., submitted).

POLYMORPHISM KpnI detects a 2 allele polymorphism with bands at 6.5 kb and 5.8 kb.

FREQUENCY Estimated from 91 unrelated CEPH Caucasians
KpnI 6.5kb: 0.37
 5.8kb: 0.63

NOT POLYMORPHIC FOR PvuII, SacI, StuI, HindIII, XhoI, AvaI, AccI, AvaII, Eco109, BglII, BglI, HincII, MspI, StyI, HinfI, MboII, RsaI, Sau3A, SacI, SspI, TaqI.

CHROMOSOMAL LOCALISATION 17p13 (G.I. Bell, submitted)

MENDELIAN INHERITANCE Co-dominant inheritance has been observed in ten families consisting of CEPH individuals.

PROBE AVAILABILITY Contact G.I. Bell.

OTHER COMMENTS RFLPs were observed under normal conditions of hybridization and washing.

REFERENCE

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