A three allele insertion polymorphism is identified by the human chromosome 19q13.3 probe pKBE0.8 (D19S119)

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Source/Description: The probe pKBE0.8 is a 0.8 kb EcoRI/BamHI fragment isolated from a human chromosome 19 cosmid library and subcloned in pSP65. pKBE0.8 contains human DNA sequences which are homologous to a genomic region approximately 240 kb telomeric to pE0.8 (Shutler *et al.*, 1991).

Polymorphism: A three allele insertion polymorphism is detected using any one of the following restriction endonucleases: BamHI (fragment sizes 4.0 kb, 3.5 kb, 3.0 kb), PstI (7.3/6.8/6.3 kb), SstI (3.7/3.2/2.7 kb), SmaI (3.9/3.4/2.9 kb), PvuII (5.5/5.0/4.5).

Not Polymorphic For: AvaII, BanI, BglI, BglII, EcoRI, HindIII, MspI, RsaI, TaqI.

Frequency: Estimated from 355 unrelated individuals
A1 = 0.58 (large fragment)
A2 = 0.27 (intermediate fragment)
A3 = 0.15 (small fragment)

Chromosome Localization: pKBE0.8 maps to human chromosome 19q13.3 distal to pE0.8 (Shutler *et al.*, 1991). The physical linkage of pKBE0.8 with pE0.8 is based upon the isolation of contiguous DNA sequences derived from a chromosome walk in a human genomic cosmid library.

Mendelian Inheritance: A codominant segregation pattern was observed in over 100 myotonic dystrophy (DM) families.

Probe Availability: The probe is available for collaborative studies on DM. It is freely available for all other studies (contact R.G.K.).

Other Comments: Close linkage is observed between pKBE0.8 and the myotonic dystrophy locus ($Z_{max} > 20.0$, $\theta_{max} = 0.00$). HincII identifies a two allele polymorphism which shows strong linkage disequilibium with the other RFLPs but is less informative in our DM population (fragment sizes 16 kb and 10 kb; frequencies 0.85/0.15). RFLPs are observed under normal hybridization and wash stringencies.

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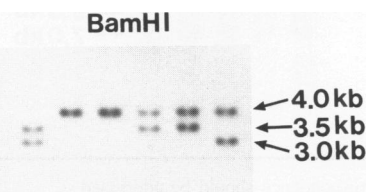
Reference: Shutler *et al.* (1991) *Genomics* **9**, 500–504.



A frequent HincII polymorphism identified by the human chromosome 19q13.3 probe pKEX0.8 (D19S118)

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Source/Description: The probe pKEX0.8 is a 0.8 kb EcoRI/XbaI genomic fragment subcloned in Bluescript (Strategene). It was isolated from a human chromosome 19 cosmid library during a chromosome walk initiated in a telomeric direction from pE0.8 (Shutler *et al.*, 1991) near the human excision repair gene ERCC1. pKEX0.8 is homologous to a chromosome 19 sequence which is located approximately 180 kb telomeric to pE0.8.

Polymorphism: HincII identifies a two allele polymorphism (fragment sizes 15.5 kb and 11.5 kb).

Not Polymorphic For: BclI, BglI, BglII, BstEII, EcoRI, HindIII, MspI, SacI, StuI.

Frequency: Estimated from 219 unrelated individuals
A1 = 0.46 (large fragment)
A2 = 0.54

Chromosome Localization: The probe pKEX0.8 is localized to human chromosome 19q13.3 distal to pE0.8 (Shutler *et al.*, 1991).

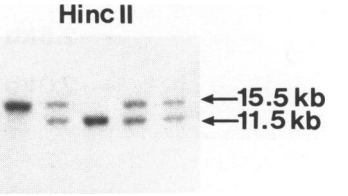
Mendelian Inheritance: A codominant segregation pattern was observed in over 100 myotonic dystrophy (DM) families.

Probe Availability: Available for collaborative studies in myotonic dystrophy. Freely available for all other studies (contact R.G.K.).

Other Comments: pKEX0.8 is closely linked to the myotonic dystrophy disease locus ($Z_{max} > 20.0$, $\theta_{max} = 0.01$). RFLPs are observed under the following wash conditions: 0.2×SSC, 0.1% SDS, 55°C.

Acknowledgements: This work was supported by grants to R.G.K. from the Medical Research Council of Canada and the Muscular Dystrophy Associations of Canada and the United States. The cosmid library from which the probe was isolated was kindly provided by Dr. Pieter de Jong, Lawrence Livermore Laboratories, Livermore, California.

Reference: Shutler *et al.* (1991) *Genomics* **9**, 500–504.



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