RFLP identified by the probe pKE0.6 (D19S117) at human chromosome 19q13.3

G.Shutler, C.Tsilfidis, S.Leblond and R.G.Korneluk*
Molecular Genetics Laboratory, Children's Hospital of Eastern Ontario, 401 Smyth Road, Ottawa K1H 8L1, Canada,

Source/Description: The probe pKE0.6 is a 0.6 kb EcoRI genomic fragment subcloned in pUC13. It was isolated from a human chromosome 19 cosmid library and is homologous to a sequence located approximately 100 kb telomeric to pE0.8 (Shutler *et al.*, 1991).

Polymorphism: PstI identifies a two allele polymorphism (fragment sizes 2.5 and 2.0 kb).

Not Polymorphic For: BamHI, EcoRI, HindIII, PvuII, SacI, TaqI.

Frequency: Estimated from 161 unrelated individuals
A1 = 0.12 (large fragment)
A2 = 0.88

Chromosome Localization: The probe pKE0.6 maps to 19q13.3 distal to pE0.8. The physical linkage of pKE0.6 with pE0.8 is based upon the isolation of contiguous DNA sequences derived from a chromosome walk in a human genomic cosmid library.

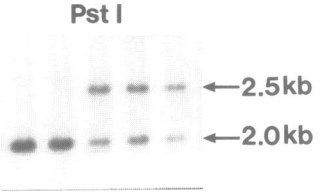
Mendelian Inheritance: A codominant segregation pattern was observed in over 100 myotonic dystrophy (DM) families.

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 between pKE0.6 and the DM locus has been observed ($Z_{max} > 10.0$, $\theta_{max} = 0.00$). The RFLP is observed under normal hybridization and wash conditions.

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.



* To whom correspondence should be addressed

An SstI RFLP detected by the probe pKE2.1 (D19S116) localized to human chromosome 19q13.3

C.Tsilfidis, G.Shutler, S.Leblond and R.G.Korneluk*
Molecular Genetics Laboratory, Children's Hospital of Eastern Ontario, 401 Smyth Road, Ottawa K1H 8K1, Canada

Source/Description: The probe pKE2.1 is a 2.1 kb EcoRI fragment isolated from a human chromosome 19 cosmid library and subcloned in pUC13. It is homologous to chromosome 19 sequences located approximately 80 kb telomeric to pE0.8 (Shutler *et al.*, 1991).

Polymorphism: SstI (SacI) identifies a two allele polymorphism with fragment sizes at 9.0 kb and 7.0 kb.

Not Polymorphic For: AlwNI, ApaI, ApaLI, AvaII, BanI, BglI, BglII, Bsu36I, EcoRI, EcoRV, HincII, HindIII, NcoI, NsiI, PstI, PvuII, RsaI, StuI, TaqI, XbaI, XmnI.

Frequency: Estimated from 378 unrelated individuals for
SstI:A1 = 0.21 (large fragment size)
A2 = 0.79

Chromosome Localization: The probe pKE2.1 is located distal to the locus detected by the probe pE0.8. The physical linkage of pEK2.1 with pE0.8 is based upon the isolation of contiguous DNA sequences derived from a chromosome walk in a human genomic cosmid library.

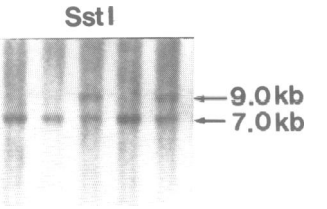
Mendelian Inheritance: A codominant segregation pattern has been observed in over 100 myotonic dystrophy (DM) families.

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

Other Comments: The polymorphism is closely linked to the myotonic dystrophy locus ($Z_{max} > 10.0$, $\theta_{max} = 0.00$). The enzyme NcoI identifies a polymorphism (fragment sizes 2.2 kb and 0.8 kb) which shows strong linkage disequilibrium with the SstI polymorphism. RFLPs are observed under normal hybridization and wash stringencies.

Acknowledgements: This work was supported by grants to R.G.K. from the Muscular Dystrophy Associations of Canada and the United States and from the Medical Research Council of Canada. The cosmid library from which pKE2.1 was isolated was kindly provided by Dr. Pieter de Jong, Lawrence Livermore National Laboratories, Livermore, California.

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



* To whom correspondence should be addressed