A BanI RFLP at a deletion hotspot in the human dystrophin gene

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Source/description: Cf56a is a 0.9 kb EcoRI fragment of dystrophin cDNA in pUC13 (1). Cf56a is identical to Kunkel's cDNA probe 8.

Polymorphism: Constant bands of 14.4, 11.0, 8.1, 6.2 and 1.3 kb correspond to exons I, N, L, M and K respectively of ref (2). The polymorphic band is exon J (exon 48, 1.2+3.9 kb HindIII bands).

Frequency: Estimated from 149 chromosomes in caucasian British

Allele 1	6.7 kb	0.62
Allele 2	6.4 kb	0.25
Allele 3	4.2 kb	0.13

Chromosomal location: Xp21.

Mendelian inheritance: Codominant segregation of all alleles in a large 4-generation family and 10 nuclear families.

Probe availability: Contact Kay Davies, Molecular Medicine, John Radcliffe Hospital, Oxford OX3 9DU (or ATCC for probe 8).

Other comments: This exon is deleted in 25% of all Duchenne/Becker dystrophy boys (2). Therefore this RFLP is useful for determining carrier status of at-risk females by showing heterozygosity or apparent non-maternity.

Acknowledgement: We thank Kay Davies for the Cf56a probe.

References: 1. Forrest, S.M., Cross, G.S., Flint, T., Speer, A., Robson, K.J.H. and Davies, K.E. (1988) *Genomics* 2, 109–114. 2. Read, A.P., Mountford, R.C., Forrest, S.M., Kenwick, S.J., Davies, K.E. and Harris, R. (1988) *Hum. Genet.* 80, 152–156.

An AluI RFLP detected in the human prion protein (PrP) gene

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Source/description: Probe pEA974(1), contains a 974 bp fragment present in Prp Clone XIV inserted into pBR322. A PvuII RFLP has been previously described (2).

Polymorphism: AluI identifies a two allele polymorphism of either a band at 965 bp, a band at 565 bp, or two bands at 965 bp and 565 bp.

Frequency: Studied in 14 European Caucasians:

965 bp allele (A1): 0.11

565 bp allele (A2): 0.89

Not polymorphic for: (studied in 14 unrelated European Caucasians)—A total of 46 enzymes have been studied including ApaI, ApaLI, AvaI, AvuI, BalI, BalII, BanI, BamHI, BclI, BglI, and BglII. A complete list of enzymes is available on request.

Chromosomal localisation: the PrP gene has been sublocalized to the p arm of chromosome 20 (3).

Mendelian inheritance: Mendelian pattern of inheritance was shown in an informative family.

Probe availability: Contact Nikolaos K. Robakis.

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References: (1) Robakis *et al.* (1986) *PNAS* 83, 6377–6381. (2) Wu *et al.* (1987) *Nucl. Acids Res.* 15, 3191. (3) Robakis *et al.* (1986) *BBRC* 140, 758–765.