RFLP for Duchenne muscular dystrophy cDNA clone 30-2

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SOURCE AND DESCRIPTION: 30-2 is one of 6 cDNA clones which comprise the cDNA for the Duchenne muscular dystrophy gene (kindly supplied by L.M. Kunkel). It is a 1.15kb fragment in the EcoRI site of Bluescribe.

POLYMORPHISM: Taq I (T[↓]CGA) identifies two bands with alleles at 3.7 and 3.5kb, as well as eight constant bands at 9.0, 7.5, 4.6, 3.6, 3.4, 2.5, 1.7 and 1.4kb.

FREQUENCY: Studied in 47 unrelated DMD males.
3.7kb allele 0.45
3.5kb allele 0.55

NOT POLYMORPHIC FOR: Hind III

CHROMOSOME LOCATION: Xp21.2, locus DMD.

MENDELIAN INHERITANCE: Co-dominant X-linked segregation demonstrated in two 2-generation families.

PROBE AVAILABILITY: Probe available from ATCC (57668, 57669).

OTHER COMMENTS: 1.1% agarose gels required to resolve the bands. The polymorphism is also recognized by PERT 87-15.

REFERENCE: M. Koenig, E.P. Hoffman, C.J. Bertelson, A.P. Monaco, C. Feener and L.M. Kunkel. Complete cloning of the Duchenne Muscular Dystrophy (DMD) cDNA and preliminary genomic organization of the DMD gene in normal and affected individuals. *Cell* 50:509-517 (1987).

ACKNOWLEDGEMENTS: This work is supported by Clinical Research Grants from the MDA of America (RJB, ADR), and a Postdoctoral Fellowship (APW) from the MDA of America, a Clinical Research Grant NINCDS #19999 (ADR) and Teacher-Investigator Development Award NS00960 (TS) from the NIH and the Neuromuscular and Telethon Foundations of Western Australia (NGL).