RFLP for HindIII at the Duchenne muscular dystrophy gene

T.W.Prior, K.J.Friedman and L.M.Silverman

Departments of Pathology and Hospital Laboratories, University of North Carolina and North Carolina Memorial Hospital, Chapel Hill 27599, NC, USA

SOURCE AND DESCRIPTION: cDMD 1-2a is a 1.54kb insert in the EcoRI HindIII sites of bluescribe. It is the most 5' of the 6 cDNA clones which comprise the complete cDNA for the Duchenne muscular dystrophy gene.

POLYMORPHISM: HindIII identifies a 2 allele polymorphism with either a band at 7.5 or 8.3kb, as well as constant bands at 3.1, 3.2, 3.25, 4.2, 4.6, 8.0, 8.5, and 10.5kb.

FREQUENCY: 39 unrelated individuals were studied.

7.5kb allele 0.90

8.3kb allele 0.10

CHROMOSOME LOCATION: Xp21.2, 5' region of the Duchenne muscular dystrophy gene.

MENDELIAN INHERITANCE: Co-dominant X-linked segregation demonstrated in four families.

PROBE AVAILABILITY: Probe available from ATCC.

OTHER COMMENTS: Absence of the 7.5kb allele may be misinterpreted as a deletion due to poor resolution of the 8.3kb allele from the constant 8.0 and 8.5kb alleles on 0.8% agarose gels. 0.6% agarose gels are required to resolve the bands.

REFERENCE: Koenig et al. Cell 50 (1987) 509-517.