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DNA amplification of a further exon of Duchenne muscular dystrophy locus increase possibilities for deletion screening

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The data of Chamberlain et al (1) allow the detection of 76% of deletions in the region Cf56a/Cf23a identified by hybridization in our patients. We have generated two oligonucleotides allowing the amplification of a further exon which is included in the 3.4kb hybridization fragment of Cf56a. This exon is deleted in about 10% of our patients.

5'cccaaaatattttagCTCCTACTCAGACTGTTACTCTGGTGACACAACCTGTGGTTACT
AAGGAAACTGCCATCTCCAAACTAGAAATGCCATCTTCCTTGATGTTGGAGGTACCTGCTCTGGC
AGATTTCAACCGGGCTTGGACAGAACTTACCGACTGGCTTTCTCTGCTTGATCAAGTTATAAAAT
CACAGAGGGTGATGGTGGGTGACCTTGAGGATATCAACGAGATGATCATCAAGCAGAAGgtatga
gaaaaaatgataaaagttggcagaagtttttctttaaaatgaagattttccaccaatcactttac
tctcctagaccattgcccaccagttcttaggcaactgtttctctctcagcaaacacattactctc
actattcagcctaagtataatcaag 3'

Fig. 1: Nucleotide sequence of the exon included in the 3.4kb hybridization fragment of Cf56a and a part of the flanking introns. Exon sequences are in upper case, and intron sequences are in lower case. Primer sequences are underlined.

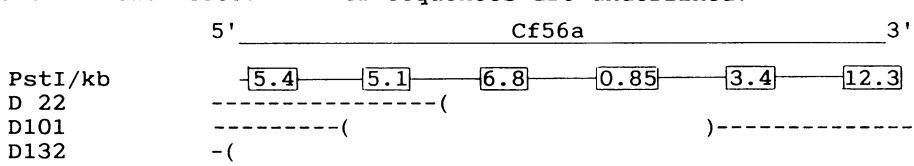
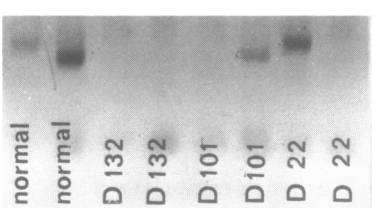


Fig. 2: Exon-containing PstI fragments detected by Cf56a and their deletion in patients D22, D101 and D132.



506bp
409bp Fig. 3: Detection of DNA amplification products of 409 bp (PstI-3.4 kb/Cf56a) and 506 bp (HindIII-1.2+3.8 kb/probe 8, equivalent of Cf56a) (1) in the deletion cases and one normal individual.

REFERENCES

1. Chamberlain, J.S. et al. (1989) Nucl Acids Research, 16 (23): 11141-11156.