Duchenne muscular dystrophy (DMD) gene cDNA 8 PstI and TaqI polymorphisms involve exon 51 of the HindIII map

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Correlation of exon-containing fragments deleted in HindIII, PstI and TaqI digests of DNA from West Australian Duchenne and Becker muscular dystrophy patients with varying deletions revealed by DMD cDNA 8 (ATCC) has demonstrated that the same exon is involved in the two polymorphisms. (One patient was deleted solely for the PstI/TaqI polymorphic exon). The exon is exon 51 as defined in the HindIII map of the gene (Koenig *et al.*, 1989). This confirms the position of the polymorphisms in TaqI (Walker *et al.*, 1989) and PstI (Gillard *et al.*, 1989) maps of the DMD cDNA 8 region. The TaqI and PstI polymorphisms are not individually highly informative, with heterozygosities of 0.18 (Laing *et al.*, 1988) and 0.21 (Darras and Francke, 1988) respectively. However, none of the 53 males studied in the present series had both the TaqI and PstI less frequent alleles. There was no evidence of linkage disequilibrium, but the rarity of the less frequent alleles would require the study of a very large number of individuals for it to be apparent. The heterozygosity of the two polymorphisms may therefore be approximately summed to give a combined heterozygosity of close to 0.39. This total heterozygosity makes the exon 51 polymorphisms more useful for carrier status diagnosis by non-inheritance of parental alleles or heterozygosity (Bartlett *et al.*, 1987). In the West Australian

population of 60 unrelated families studied with DMD cDNA probes, 6 out of 38 deletions included the TaqI/PstI polymorphic exon. Therefore, carrier status may be determined in 16% of deletion families using the TaqI and PstI polymorphisms, and heterozygosity can be expected in greater than one in three women if both polymorphisms are studied.

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