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Screening of macrodeletions of Duchenne Muscular Dystrophy (DMD) patients in Sri Lankan population by a multiplex PCR system

Duchenne Muscular Dystrophy (DMD) is an z-linked recessive disorder resulting in progressive degeneration of muscle. DMD is the most common lethal x-linked disease in the human that affects 1 in 3500 live male births, which has led to an emphasis of the mutation analysis/genetic basis of DMD patients.

Of the mutations, 60% are macrodeletions, 6% are duplications and the rest account for point mutations, microdeletions and microinsertions. One third of all the mutations arises from new mutations and two third are familial.

The deletion mutations of the gene are clustered in two High Frequency Deletion Regions (HFDRs) located in the 5' terminus and more central 3'terminus. Multiplex PCR system amplifies 18 exons, 7 exons of the 5' HFDR and 11 exons of the 3' HFDR carried out in two separate reactions. A multicentre study has verified the reliability of this multiplex PCR system which detects 98% of deletion detectable by southern CDNA hybridization.

We report a study of 30 clinically evaluated patients analysed by this multiplex PCR system, which amplifies 18 exons located in the two HFDRs. The system detected at least one deletion in 255 patients (83%). The percentage of deletions observed is higher than reported elsewhere in the world (65-70%). Exons 45, 47, 48, 50 were deleted in one third of all patients and had deletion frequencies of 14%, 15%, 18% and 14% respectively, of all the deletions.