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Biochemistry, Cytogenetics and *DMD* Gene Mutations in South Indian Patients with Duchenne Muscular Dystrophy

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ABSTRACT Thirty children aged 3-10 years with clinically confirmed or suspected Duchenne Muscular Dystrophy (DMD) were analyzed for chromosomal aberrations using cytological preparations, biochemical changes using enzyme kit protocol, and deletions in the 26 exons of the *DMD* gene by targeting the mutations at the proximal and distal 'hot spot' regions of the dystrophin gene in South Indian patients with DMD. The frequency of chromosomal aberrations (both chromosomal and chromatid-type) and serum enzyme levels were significantly elevated in DMD subjects as compared to controls. Multiplex PCR assays revealed 27 patients having deletions in the *DMD* gene located at the distal 'hot spot' region. This study suggests that disease progression is directly associated with higher incidence of the deletions at the distal 'hot spot' of the *DMD* gene.