DELETION SCREENING OF SRI LANKAN DUCHENNE MUSCULAR DYSTROPHY PATIENTS USING THE POLYMERASE CHAIN REACTION

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Duchenne Muscular Dystrophy (DMD) is a severe X-chromosome linke myopathy caused by a defect in the DMD gene which is located i the Xp21 band. The DMD gene is a large one of approximately 200 Kb consisting of not less than 65 exons, and codes for a 400 k prodein called Dystrophin. The large size of the gene makes i deletion prone with a known deletion frequency of 66%.

Our study has the objectives of determining whether the deletio frequency and the regions deleted of our DMD population is th same as that found elsewhere. Here we report the results of thi study using a group of 24 DMD patients (Sri Lankan) employing th technique of multiplex DNA amplification using the Polymeras chain reaction (1). Six sets of oligonucleotides from the DM gene were used as primers for amplification. Our result indicated that 62.5% of the patients contained deletions in th DMD gene, a result which is in agreement with values obtained i similar studies carried out elsewhere in the world. The result also indicated that 80% of the deletions were localized in region of the DMD gene now regarded as a "Hot Spot" for Duchenn deletions.

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References: 1. Jeffrey S Chamberlain et al (1988).
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