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Research Title : *Mutation analysis in Saudi Duchenne and Becker muscular dystrophy patients using multiplex PCR*

Mutation analysis in Saudi Duchenne and Becker muscular dystrophy patients using multiplex PCR

Descriptipn : Introduction: In Saudi Arabia, only limited work has been reported on the mutation patterns of Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD). This study looks at the spectrum of deletions in the hot spot regions of the DMD gene in Saudi DMD/BMD patients using an enhanced multiplex PCR technique. Material and methods: Twenty-six exons of the DMD gene were analyzed, in eight unrelated DMD/BMD cases aged 4-19 years, using four different multiplex PCR sets. Each multiplex PCR set amplified a total of six or seven exons. Normal controls were included for validation. Results: Using an optimized multiplex PCR method, 5 out of 8 DMD/BMD patients showed deletions, while the remaining three had no deletions in regions analyzed. Set 1 detected no deletions in any of the patients, whereas each of sets 2, 3 and 4 detected two, four and three deletions respectively. All of these mutations were located in the distal 'hot spot' region. No deletions were detected in the proximal 'hot spot' region. The normal control samples showed no deletions in any of the 26 exons tested. Conclusions: In this study, multiplex PCR technology was utilized to demonstrate the frequency of the most commonly found deletions in a limited group of Saudi DMD/BMD patients. The overall distribution of deletion mutations in the distal 'hot spot' region was in accordance with DMD/BMD cases investigated elsewhere. The study also serves as a good starting point for further investigations into the genetic aspects of the Saudi DMD/BMD population

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