

Spectrum of dystrophin gene mutations observed in patients suspected of Duchenne muscular dystrophy in Pakistan

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Introduction

Duchenne muscular dystrophy (DMD) is an inherited X-linked recessive genetic disorder caused by a defect in the dystrophin gene. This results in a quantitative and/or qualitative abnormality in the dystrophin protein causing progressive muscular damage and weakness.

Objective

To investigate the spectrum of deletions and duplications in the dystrophin gene of patients suspected of DMD or of being DMD carriers.

Methods

A retrospective review of DMD testing performed from March 2017 to June 2018 was conducted at Aga Khan University Hospital, Karachi. Multiplex ligation-dependent probe amplification (MLPA) was used for identification of deletion/duplication in 79 exons of the dystrophin gene.

Results

In a total of 92 cases, 85 males and 7 females were examined. In males, deletions and duplications were identified in 44 (52%) of the 85 cases; deletions were found in 38 cases (45%) and duplications in 6 cases (7%). In seven cases referred for females, two of the seven females (30%) were found to have a heterozygous deletion, suggestive of carrier status.

Conclusions

This study identifies deletions in dystrophin exon 42–52 to be the most common while duplications were mostly found in exons 3–7. This information can facilitate the use of new treatments with exon-skipping drugs which are mutation specific for DMD. ■

Conflict of interest statement

None declared.

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