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Detection of deletions and duplications in the Duchenne muscular dystrophy gene by the molecular method MLPA in the first Argentine affected families

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ABSTRACT. Deletions/duplications in the Duchenne muscular dystrophy (DMD) gene account for 60 to 70% of all alterations. A new technique, multiplex ligation-dependent probe amplification (MLPA), has been described that allows the detection of large genetic rearrangements by simultaneous amplification of up to 45 target sequences. The present article is based on the diagnosis of the first Argentine affected families by the application of MLPA. DNA samples from patients with and without a previous diagnosis were included. MLPA assays were performed according to manufacturer recommendations. Polymerase chain reaction and direct sequencing were performed when a single-exon deletion was detected. Results

were analyzed using the Gene Marker v1.6 and Sequencing Analysis v5.2 software. In the samples with a previous diagnosis (as identified by short tandem repeat-polymerase chain reaction analysis), MLPA confirmed in some samples the same deletion and detected in others a larger deleted fragment. This enabled the prediction of the expected male phenotype. One deletion and one duplication were detected in patients without previous diagnosis. In this study, we investigated the applicability of MLPA in our country. Our results showed a 100% confirmation of the deleted fragments detected by short tandem repeat segregation analysis. Moreover, in some cases, the MLPA assay was able to refine the breakpoints involved. In addition, MLPA identified deletions/duplications in samples without previous diagnosis. In comparison to the available diagnosis strategies in Argentina, MLPA is less time-consuming, and spans the complete coding region of DMD. The application of MLPA will improve the genetic diagnosis of DMD/Becker muscular dystrophy in our country.

Key words: Duchenne muscular dystrophy; Becker muscular dystrophy; Deletions/duplications; Multiplex ligation-dependent probe amplification