

SMALL MUTATIONS OF THE DYSTROPHIN GENE

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an intermediate RPR. This could be explained by the fact that the deletion created a novel sequence similar to the probe-hybridizing sequence, thereby inducing competition between two sequences for probe hybridization. This appears to be a new mechanism that can disturb the MLPA reactions.

An intermediate grade of amplified product is described here for the first time in two Japanese patients (Figs. 2 and 3). We suspect that ambiguous amplification results shown in this study have been dismissed as artifacts in previous studies without further examination. Therefore, we recommend that detailed examination of ambiguous MLPA results should be conducted, especially in patients with genotype-phenotype mismatch.

In conclusion, MLPA is a powerful tool for screening not only exon deletions and duplications, but also small mutations in the dystrophin gene, and could be used in routine clinical diagnosis.

AUI ► Disclosure Statement

No competing financial interests exist.

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AUTHOR QUERY FOR GTMB-2009-0002-OKIZUKA_1P

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