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Splicing mutation of a gene within the Duchenne muscular dystrophy family

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ABSTRACT. The aim of this study was to identify the mutation site and phenotype of the Duchenne muscular dystrophy (*DMD*) gene in a DMD family. The *DMD* gene is by far the largest known gene in humans. Up to 34% of the point mutations reported to date affect splice sites of the *DMD* gene. However, no hotspot mutation has been reported. Capture sequencing of second-generation exons was used to investigate the *DMD* gene in a proband. Sanger sequencing was performed for mutation scanning in eight family members. Scale-invariant feature transform and PolyPhen were applied to predict the functional impact of protein mutations. A hemizygous splicing mutation IVS44ds +1G-A (c.6438 +1G>A) that induces abnormal splicing variants during late transcription and produces abnormal proteins was located in intron 44. Four missense mutations (p.Arg2937Gln, p.Asp882Gly, p.Lys2366Gln, and p.Arg1745His) that are known multiple-polymorphic sites were found in the coding region of the *DMD* gene. A heterozygous c.6438

+1G>A mutation was detected on the X chromosome of the proband's mother and maternal grandmother.

Key words: Capture sequencing of second-generation exons; Duchenne muscular dystrophy; Gene; Mutation; Pedigree study

