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Identification of *DMD* Mutation in Korean Siblings Using Full Gene Sequencing

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ABSTRACT Duchenne Muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutations in the dystrophin gene, which is located in Xp21. The majority of the identified mutations are large deletions and duplications, and gene dosage assays were developed for quantitative genomic screening of copy number variations. However, remaining 25% of the DMD are due to point mutations and require direct full gene sequencing. We report Korean siblings with novel small intragenic duplication in an exon 41 (c.5756dupT) which was detected by direct sequencing of whole dystrophin (*DMD)* gene exons. This 1-bp duplication is a novel frameshift mutation and induces premature termination (p.Leu1919Phefs*13). Gene therapy in DMD has been developed and it is important to know the exact mutation site and type to predict prognosis and to prepare further therapy. Therefore, in DMD patients with normal *DMD* gene dosage, direct sequencing of *DMD* gene is essential to detect small intragenic deletions/insertions and missense, nonsense, and splicing mutations.