DMD Dystrophin Gene Testing | LeXDMD Research Panel

Background

The *DMD* gene, located on theX chromosome (Xp21.2), encodes dystrophin, spanning over 2.2 Mb and accounting for about 0.1% of the human genome or 1.5% ofthe X chromosome. 99% of its sequence consists of intronic regions, while its 79 exons comprise only about 1%. Owing to its super-long sequence and complex exon structure, the mutation rate ofthe *DMD* gene is considerably higher than that of other genes associated with single-gene disorders. Complete or partial loss of function of this gene can lead to severe genetic disorders such as Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD).

In contrast to traditional stepwise testing, full-length sequencing of the *DMD* gene can precisely detect pathogenic deep-intronic variants, structural variations, and complex rearrangements, providing key technical support for elucidating molecular patho-genesis and formulating personalized treatment strategies. Early and precise diagnosis is not only fundamental to delaying disease progression, guiding corticosteroid therapy, and optimizing family planning but also plays a profound role in improving patients' quality of life.

Introduction

LeXDMD Research Panel v1.0 targets the *DMD* gene that encodes dystrophin, covering an approximately 2.2 Mb genomic region for enrichment and comprehensive analysis of the entire *DMD* gene sequence.

Performance

Basic Quality Control Performance on Dual Platforms

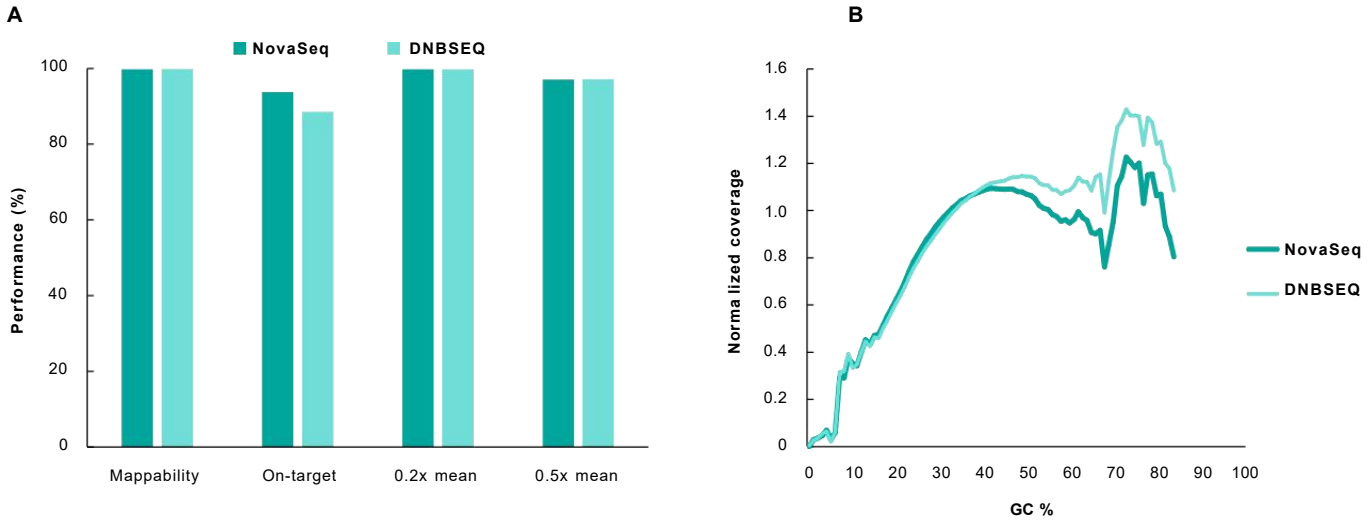

Figure 1. Basic quality control performance ofthe LeXDMD Research Panel v1.0. A. Mappability, On-target rate, and Target covered; **B.** GC bias. Pre-library prep- aration was performed using the LeXPrep EZ DNA Library Preparation Kit with the LeXPrep Universal Stubby Adapter (UDI) Module, followed by hybrid capture using the LeXDMD Research Panel v1.0 and LeXPrep Hybrid Capture Reagents. Sequencing was performed on NovaSeq 6000 (PE150) and DNBSEQ-T7 (PE150).

Note: Samples were human genomic DNA standard (Promega, G1471).

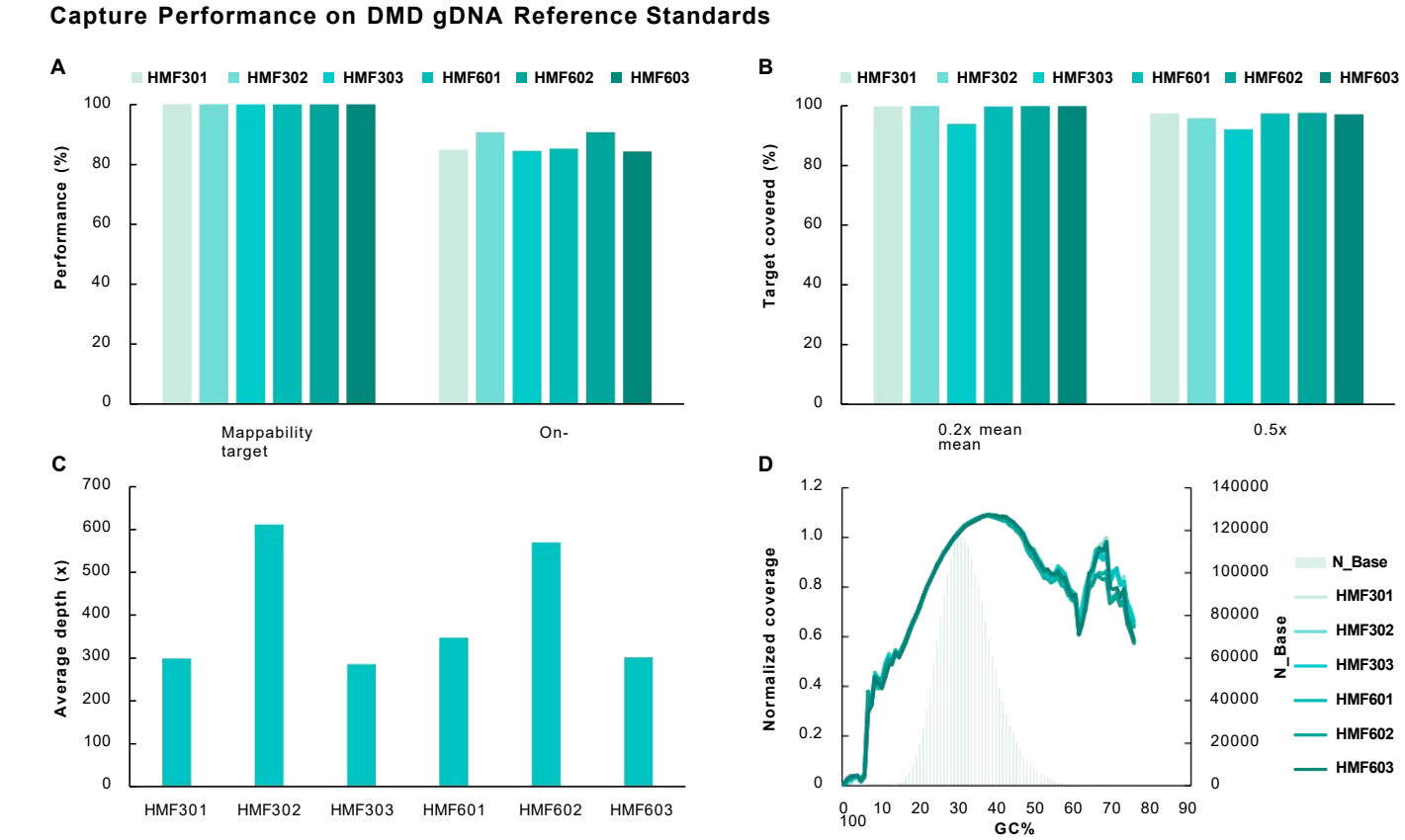

Figure 2. Capture performance of the LeXDMD Research Panel v1.0 on reference standards. A. Mappability & On-target rate; **B.** Target covered; **C.** Average sequencing depth (without deduplication); **D.** GC bias. Sequencing was performed on NovaSeq 6000 (PE150). Data of 0.78, 1.5, 0.75, 0.9, 1.4, and 0.79 Gb were used for analysis.

Note: Samples are derived from DMD gDNA Reference Standards (GeneWell). HMF301-3 correspond to GW-HMF301-3, and HMF601-3 correspond to GW-HMF601-3; within the two families, the gender of standards 1-3 are Male, Female, and Male, respectively; Genotypes of HMF301-3 by MLPA are Normal, Exon48-Exon50 hetero- zygous deletion and Exon48-Exon50 deletion/hemizygote, while genotypes for HMF601-3 are Normal, Exon18-Exon25 haplox repeat, and Exon18-Exon25 repeat.

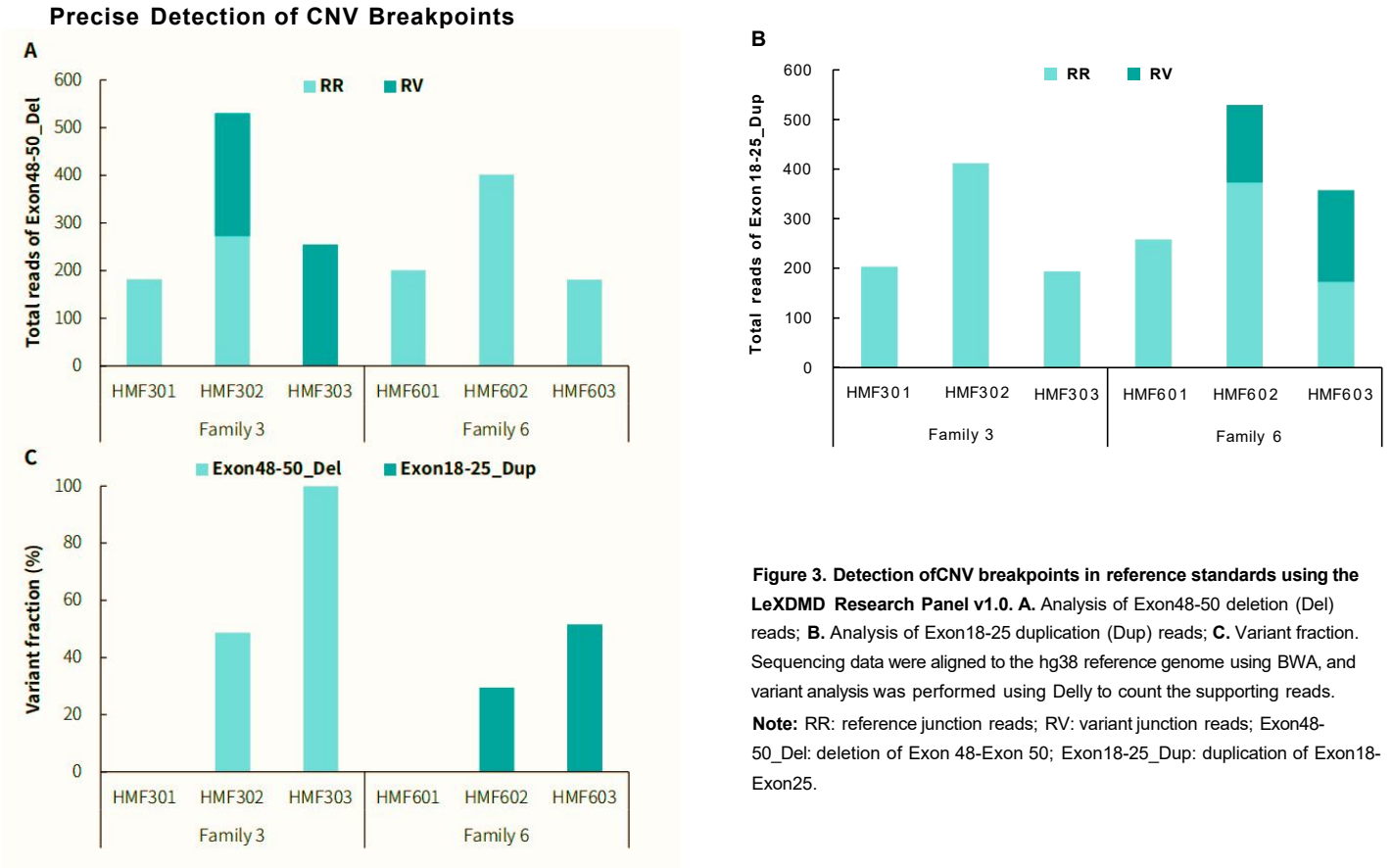

Figure 3. Detection ofCNV breakpoints in reference standards using the LeXDMD Research Panel v1.0. A. Analysis of Exon48-50 deletion (Del) reads; **B.** Analysis of Exon18-25 duplication (Dup) reads; **C.** Variant fraction. Sequencing data were aligned to the hg38 reference genome using BWA, and variant analysis was performed using Delly to count the supporting reads.

Note: RR: reference junction reads; RV: variant junction reads; Exon48-50_Del: deletion of Exon 48-Exon 50; Exon18-25_Dup: duplication of Exon18-Exon25.

DMD Genomic Coverage

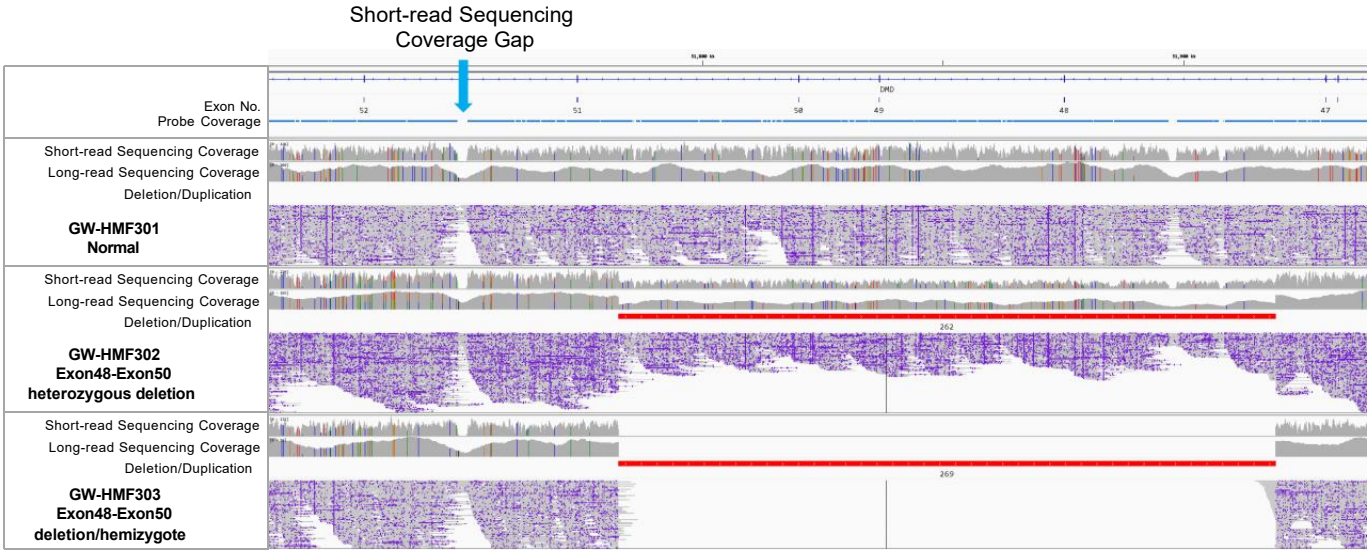

Figure 4. Genomic coverage of DMD gDNA Reference Standards after targeted capture with the LeXDMD Research Panel v1.0, as assessed by short-read (NovaSeq 6000, PE150) and long-read (Oxford Nanopore) sequencing.

Ordering Information

Product	Scale	Catalog#
LeXDMD Research Panel v1.0, 16 rxn	16 rxn	LX01892
LeXDMD Research Panel v1.0, 96 rxn	96 rxn	LX01891

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