Diagnostic Genome Sequencing Test Report

Personal Information

Name: John Doe
Relation: -
Sex/Birth: M / 0000-00-00

Specimen Information

Sample ID: 20260101-000-0000
Medical record No: 012345
Date received: 2026-01-01

Test Information

Test reported: 2026-02-11
Ordering physician: -
Institution: GC Genome hospital

TEST PERFORMED

DGS (Diagnostic Genome Sequencing)

CLINICAL INFORMATION

Clinical weakness of limbs: increased GOT, increased GPT, increased CK, calf hypertrophy, gower (+), normal echocardiogram

RESULT

POSITIVE

A hemizygous pathogenic variant was identified in the DMD gene.

Gene	DNA change	Predicted AA change	Zygosity	OMIM Disease	Inherit	Class
DMD	c.10210dup	p.Asp3404GlyfsTer29	Hem	DMD/BMD	XLR	PV

Reference sequence: NM_004006.3(DMD)
OMIM disease: DMD/BMD, Duchenne/Becker muscular dystrophy
Abbreviation: Hem, Hemizygous; PV, Pathogenic Variant; XLR, X-linked recessive

INTERPRETATION

[2025.11.24]
DMD, c.10210dup (p.Asp3404GlyfsTer29)
This variant changes reading frame of the DMD gene and the 29th amino acid to a stop codon due to the 3404th amino acid, Aspartic acid, is substituted with Glycine. This variant is not present in population databases. This variant has been reported in individuals affected with Muscular dystrophy (PMID: 28116794). ClinVar contains an entry for this variant as Pathogenic (Variation ID: 931458) For these reasons, this variant has been classified as Pathogenic.

Pathogenic DMD variants are associated with Duchenne/Becker muscular dystrophy.

RECOMMENDATIONS

Clinical correlation and familial testing is recommended if necessary.

INCIDENTAL FINDINGS

No (Likely) Pathogenic Variant was identified in the 84 genes recommended by ACMG
* Investigation of 84 genes recommended by ACMG SF v3.3 (Genet Med. 2025.)

Tested by: Hae-In Ryu M.T.(51943) Reported by/Reviewed by: Chang-Ahn Seol M.D.(1037) Sae-Mi Lee M.D.(1067)

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METHODS

Genomic DNA was extracted from EDTA whole blood and sequenced with paired-end reads on Illumina NovaSeq 6000 system. The DNA sequence reads were aligned to reference sequence based on public human genome build GRCh37/UCSC hg19. Using a in-house bioinformatics pipeline, data were filtered and analysed to identify sequence variants. CNV calling is based on parliament2 pipeline.

Evaluation is focused on coding exons along with flanking +/-20 intronic bases, however extended to the complete gene region for candidate genes or in search for a second previously described variant in autosomal recessive inheritance pattern. Sequence variants were classified based on the ACMG/AMP guidelines (Richards et al., 2015). Reported results are focused on pathogenic and likely pathogenic variants in genes related to the phenotype of proband, while variants of uncertain significance are only rarely reported at our discretion. Variants that pass internal QC criteria are not validated by Sanger sequencing.

ANALYSIS STATISTICS

Mean depth of coverage	113.23X
% of > 10x	98.66%

LIMITATIONS

The absence of definitive pathogenic findings does not rule out the diagnosis of a genetic disorder as some genetic abnormalities may be undetectable with this test. It is possible that the genomic region where a disease-causing variant exists in the proband was not captured or sufficiently sequenced with low quality. Additionally, multifactorial disorders and some types of genetic disorders due to nucleotide repeat expansion/contraction, abnormal DNA methylation, and other mechanisms may not be detectable with this test. This test also cannot reliably detect mosaicism, chromosomal aberrations, and deletions/insertions of 20 bp or more. Some genes have inherent sequence properties (for example: repeats, homology, high GC content, rare polymorphisms) that may result in suboptimal data, and variants in those regions may not be reliably identified.

※ This test was developed and its performance characteristics determined by GC Genome. It has not been cleared or approved by the Korean Ministry of Food and Drug Safety (MFDS).

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