CENTER FOR ANATOMY AND CELL BIOLOGY
Medical University of Vienna

Neuromuscular Research (NMRD)
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EXOME SEQUENCING

DIAGNOSTIC REPORT

Klinik Favoriten
Department of Pediatrics and Adolescent Medicine
Dr. [redacted]
Kundratstraße 3, 1100 Vienna

Vienna, December 2, 2025

Patient Name:	**FISCHER, Constantin** (Male)	**Date of Birth:**	March 17, 2019
Clinical Diagnosis:	**Duchenne Muscular Dystrophy** (trunk-predominant muscle weakness, Gowers' sign, calf hypertrophy, serum CK > 20,000 U/L)		
Previous Findings:	Unremarkable MLPA result (our report dated October 25, 2025)		
Test Requested:	Whole-Exome Sequencing (Order: Oct 27, 2025)	**Specimen:**	EDTA Blood (Received: Oct 10, 2025)

MOLECULAR GENETIC RESULTS AND INTERPRETATION

By means of next-generation sequencing and direct DNA sequencing, a hemizygous single-nucleotide duplication was detected in exon 27 of the *DMD* gene ("Dystrophin"):

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DMD:
NM_004[redacted]39Glyfs*3) hemizygous
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In the scientific literature, pathogenic variants in the *DMD* gene (OMIM * 300377) are well described in a causal genetic context with X-linked recessive muscular dystrophies.

The detected 1-bp insertion causes a frameshift resulting in a premature termination codon and is therefore formally classified as pathogenic; it has been previously described in the scientific literature as a pathogenic variant in patients with Duchenne muscular dystrophy (Flanigan KM et al. *Hum Mutat* 30:165, 2009; HGMD Public: C1098857). The classification according to ACMG criteria is: **Pathogenic**.

We recommend performing carrier testing for the patient's mother and/or within the extended family.

SUMMARY

The results of the molecular genetic investigations performed are compatible with the presence of an **X-linked recessive dystrophinopathy**, formally most consistent with **Duchenne type** (DMD, "Duchenne muscular dystrophy"; OMIM #310200), in full agreement with the clinically suspected diagnosis.

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