Patient name: Vasiliy Ivanov
Date of birth: 01/05/2010
Lab Accession: XD7646
Gender: Male
Race: White

Specimen type: Bloow
Date specimen received: 01/31/2014
Referring physician: John Smith, MD PhD
Referring facility: Pediatric hospital

Indication for test — Muscular dystrophy, low blood level
Test performed — Distrophynopaties
Genes tested: DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B, DMD, SCN1A, SCN1B.

Result: Positive - An established cause of the reported phenotype was identified.

Low sequencing coverage of genes were identified.

DNA variants:

Gene	ACMG classification	Zygosity	Variant	Disease
DMD	Likely pathogenic	hemi	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE
DMD	Pathogenic	hetero	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE
DMD	Unknown significance	homo	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE

Summary: Mutations in DMD genes were identified. This is linked to Duchenne muscular dystrophy.

Recommendations: We recommend hospitalization with a course of intense therapy by neurologies and genetics. The course should be repeated annually.

Limitations: Absence of a plausible explanation for the reported phenotype by next generation sequencing does not exclude a genetic basis of the patient's condition. Some types of genetic abnormalities, such as copy number changes, may not be detectable with the technologies performed by this next generation analysis test. It is possible that the genomic region where a disease causing mutation exists in the proband was not captured using the current technologies and therefore was not detected. Additionally, it is possible that a particular genetic abnormality may not be recognized as the underlying cause of the genetic disorder due to incomplete scientific knowledge about the function of all genes in the human genome and the impact of variants in those genes. Only variants in genes associated with the medical condition, or thought to be clinically relevant potentially for the proband's medical condition, are reported here.

Other DNA variants:

Gene	ACMG classification	Zygosity	Variant	Disease
DMD	Likely pathogenic	hemi	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE
DMD	Pathogenic	hetero	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE
DMD	Unknown significance	homo	c.5839G>T (p.Glu1947*)	#310200 MUSCULAR DYSTROPHY, DUCHENNE TYPE