INVITAE

INVITAE DIAGNOSTIC TESTING RESULTS

Patient name:	Vlad L Lupascu	**Sample type:**	Blood	**Report date:**	08/03/2021
DOB:	02/10/2002	**Sample collection date:**	03/17/2021	**Invitae #:**	RQ2560641
Sex:	Male	**Sample accession date:**	03/18/2021	**Clinical team:**	Laura Damian
MRN:	330041				

Reason for testing

Diagnostic test for a personal history of disease

Test performed

Sequence analysis and deletion/duplication testing of the 194 genes listed in the Genes Analyzed section.

- Invitae Hereditary Rhabdomyolysis Panel
- 142 individual genes

RE-REQUISITION REPORT: This report supersedes RQ2074090 (03.31.2021) and includes additional analyses.

RESULT: UNCERTAIN

Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
COL6A2	c.2171G>T (p.Arg724Leu)	heterozygous	Uncertain Significance
RYR1	c.14021G>A (p.Arg4674Gln)	heterozygous	Uncertain Significance
SUN1	c.1228T>G (p.Ser410Ala)	heterozygous	Uncertain Significance

About this test

This diagnostic test evaluates 194 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

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Next steps

- This test did not identify any pathogenic variants, but includes at least one result that is not completely understood at this time. Please note that the classification of variants may change over time as a result of new variant interpretation guidelines and/or new information. If an uncertain variant is reclassified, Invitae will update this report with the new interpretation and provide notification. This result should be discussed with a healthcare provider, such as a genetic counselor, to learn more about this result and the appropriate next steps for further evaluation. Clinical follow up may still be warranted. This result should be interpreted within the context of additional laboratory results, family history and clinical findings.

- Testing of up to two family members for the Variant(s) of Uncertain Significance (VUS) identified in COL6A2 and RYR1 is available at no additional cost. Please consider this individual's clinical features and availability of informative family members to test before ordering VUS resolution testing. More details on our VUS Resolution Program, including required documentation, can be found at **www.invitae.com/family**.

- Register your test at **www.invitae.com/patients** to download a digital copy of your results. You can also access educational resources about how your results can help inform your health.

Clinical summary

A Variant of Uncertain Significance, c.2171G>T (p.Arg724Leu), was identified in COL6A2.

- The COL6A2 gene is associated with autosomal dominant and recessive Bethlem myopathy 1 (BTHLM1) (MedGen UID: 331805) and Ullrich congenital muscular dystrophy 1 (UCMD1) (MedGen UID: 98046), collectively known as type VI collagenopathies (MedGen UID: 468393). Other COL6A2-related disorders have also been reported (OMIM: 120240).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- This variant qualifies for complimentary family studies as part of our VUS Resolution Program. Familial VUS testing is recommended if informative family members are available and are likely to provide additional evidence for future variant reclassification. Details on our VUS Resolution Program can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.14021G>A (p.Arg4674Gln), was identified in RYR1.

- The RYR1 gene is associated with autosomal dominant and recessive central core disease (CCD) (MedGen UID: 199773), autosomal recessive congenital myopathy with fiber-type disproportion (CFTD) (MedGen UID: 108177), and autosomal recessive multiminicore disease (MmD) (MedGen UID: 340597). It is also associated with autosomal recessive and autosomal dominant centronuclear myopathy (CNM) (MedGen UID: 808163) and malignant hyperthermia susceptibility type 1 (MHS1) (MedGen UID: 443948). The RYR1 gene also has preliminary evidence supporting a correlation with periodic paralysis (PMID: 29298851).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- This variant qualifies for complimentary family studies as part of our VUS Resolution Program. Familial VUS testing is recommended if informative family members are available and are likely to provide additional evidence for future variant reclassification. Details on our VUS Resolution Program can be found at https://www.invitae.com/family.

A Variant of Uncertain Significance, c.1228T>G (p.Ser410Ala), was identified in SUN1.

- The SUN1 gene currently has no well-established disease association; however, there is preliminary evidence supporting a correlation with autosomal recessive myoclonic atonic epilepsy (PMID: 31170314) and Emery-Dreifuss muscular dystrophy (EDMD) (PMID: 25210889).
- Not all variants present in a gene cause disease. The clinical significance of the variant(s) identified in this gene is uncertain. Until this uncertainty can be resolved, caution should be exercised before using this result to inform clinical management decisions.
- Familial VUS testing is not offered. Testing family members for this variant will not contribute evidence to allow variant reclassification. Details on our VUS Resolution and Family Variant Testing Programs can be found at https://www.invitae.com/family.

Variant details

COL6A2, Exon 26, c.2171G>T (p.Arg724Leu), heterozygous, Uncertain Significance

- This sequence change replaces arginine with leucine at codon 724 of the COL6A2 protein (p.Arg724Leu). The arginine residue is weakly conserved and there is a moderate physicochemical difference between arginine and leucine.
- This variant is present in population databases (rs145450812, ExAC 0.008%).
- This variant has been observed in an individual with clinical suspicion of limb-girdle muscular dystrophy (PMID: 30564623). ClinVar contains an entry for this variant (Variation ID: 288448).
- Algorithms developed to predict the effect of missense changes on protein structure and function (SIFT, PolyPhen-2, Align-GVGD) all suggest that this variant is likely to be tolerated, but these predictions have not been confirmed by published functional studies and their clinical significance is uncertain.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

RYR1, Exon 96, c.14021G>A (p.Arg4674Gln), heterozygous, Uncertain Significance

- This sequence change replaces arginine with glutamine at codon 4674 of the RYR1 protein (p.Arg4674Gln). The arginine residue is highly conserved and there is a small physicochemical difference between arginine and glutamine.
- This variant is not present in population databases (ExAC no frequency).
- This variant has not been reported in the literature in individuals with RYR1-related conditions.
- Advanced modeling of protein sequence and biophysical properties (such as structural, functional, and spatial information, amino acid conservation, physicochemical variation, residue mobility, and thermodynamic stability) performed at Invitae indicates that this missense variant is expected to disrupt RYR1 protein function.
- Algorithms developed to predict the effect of sequence changes on RNA splicing suggest that this variant may create or strengthen a splice site, but this prediction has not been confirmed by published transcriptional studies.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

SUN1, Exon 10, c.1228T>G (p.Ser410Ala), heterozygous, Uncertain Significance

- This sequence change replaces serine with alanine at codon 410 of the SUN1 protein (p.Ser410Ala). The serine residue is weakly conserved and there is a moderate physicochemical difference between serine and alanine.
- This variant is not present in population databases (ExAC no frequency).
- This variant has not been reported in the literature in individuals with SUN1-related conditions.
- Algorithms developed to predict the effect of missense changes on protein structure and function (SIFT, PolyPhen-2, Align-GVGD) all suggest that this variant is likely to be tolerated, but these predictions have not been confirmed by published functional studies and their clinical significance is uncertain.
- In summary, the available evidence is currently insufficient to determine the role of this variant in disease. Therefore, it has been classified as a Variant of Uncertain Significance.

Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. Results are negative unless otherwise indicated in the report. Benign and Likely Benign variants are not included in this report but are available upon request. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details.

GENE	TRANSCRIPT
ACAD9	NM_014049.4
ACADM	NM_000016.5
ACADVL	NM_000018.3
ACTA1	NM_001100.3
ADSSL1	NM_199165.2
AGL	NM_000642.2
AGRN	NM_198576.3
ALDOA	NM_000034.3
ALG14	NM_144988.3
ALG2	NM_033087.3
AMACR	NM_014324.5
AMPD1	NM_000036.2
ANO5	NM_213599.2
ATP2A1	NM_173201.3
ATP7B	NM_000053.3
B3GALNT2	NM_152490.4
B4GAT1	NM_006876.2
BAG3	NM_004281.3
BIN1	NM_139343.2
CACNA1S	NM_000069.2
CAPN3*	NM_000070.2
CASQ1	NM_001231.4
CAV3	NM_033337.2
CCDC78	NM_001031737.2
CFL2	NM_021914.7
CHAT	NM_020549.4
CHKB	NM_005198.4
CHRNA1	NM_000079.3
CHRNB1	NM_000747.2
CHRND	NM_000751.2
CHRNE	NM_000080.3
CLCN1	NM_000083.2
CNTN1	NM_001843.3
COL12A1	NM_004370.5
COL13A1	NM_001130103.1
COL6A1	NM_001848.2
COL6A2	NM_001849.3
COL6A3	NM_004369.3
COLQ	NM_005677.3
COQ8A	NM_020247.4
COQ9	NM_020312.3
CPT1A	NM_001876.3
CPT2	NM_000098.2
CRYAB	NM_001885.2
CTDP1*	NM_004715.4
DAG1	NM_004393.5
DES	NM_001927.3
DGUOK	NM_080916.2
DMD	NM_004006.2
DNAJB6	NM_058246.3
DNM2	NM_001005360.2
DOK7	NM_173660.4
DPAGT1	NM_001382.3
DPM1	NM_003859.1
DPM2	NM_003863.3
DPM3	NM_153741.1
DYSF	NM_003494.3
EMD	NM_000117.2
ENO3	NM_053013.3
ETFA	NM_000126.3
ETFB	NM_001985.2
ETFDH	NM_004453.3
FDX2	NM_001031734.3
FHL1	NM_001449.4
FKBP14	NM_017946.3
FKRP	NM_024301.4
FKTN	NM_001079802.1
FLAD1	NM_025207.4
FLNC*	NM_001458.4
GAA	NM_000152.3
GBE1	NM_000158.3
GFPT1	NM_001244710.1
GMPPB	NM_021971.2
GNE	NM_001128227.2
GOSR2	NM_004287.3
GYG1	NM_004130.3
GYS1	NM_002103.4
HACD1	NM_014241.3
HADH	NM_005327.4
HADHA	NM_000182.4
HADHB	NM_000183.2
HMBS	NM_000190.3
HNRNPA2B1	NM_031243.2
HNRNPDL	NM_031372.3
ISCU	NM_213595.3
ISPD	NM_001101426.3
ITGA7	NM_002206.2
KBTBD13	NM_001101362.2
KCNJ2	NM_000891.2
KLHL40	NM_152393.3
KLHL41	NM_006063.2
KLHL9	NM_018847.3
LAMA2	NM_000426.3
LAMB2	NM_002292.3
LAMP2	NM_002294.2
LARGE1	NM_004737.4
LDB3	NM_001080116.1;NM_001171610.1;NM_007078.3
LDHA	NM_005566.3
LIMS2	NM_001136037.2
LMNA	NM_170707.3
LMOD3	NM_198271.4
LPIN1	NM_145693.2
LRP4	NM_002334.3
MAP3K20	NM_016653.2

GENE	TRANSCRIPT
MATR3	NM_199189.2
MEGF10	NM_032446.2
MICU1	NM_006077.3
MTM1	NM_000252.2
MTMR14	NM_022485.4
MUSK	NM_005592.3
MYH2	NM_017534.5
MYH7	NM_000257.3
MYL2	NM_000432.3
MYO18B	NM_032608.6
MYOT	NM_006790.2
MYPN	NM_032578.3
NEB*	NM_001271208.1
OPA1	NM_015560.2;NM_130837.2
OPA3	NM_025136.3
ORAI1	NM_032790.3
PDSS2	NM_020381.3
PFKM	NM_000289.5
PGAM2	NM_000290.3
PGK1	NM_000291.3
PGM1*	NM_002633.2
PHKA1	NM_002637.3
PHKB	NM_000293.2;NM_001031835.2
PLEC	NM_000445.4;NM_201378.3
PNPLA2	NM_020376.3
POLG	NM_002693.2
POLG2	NM_007215.3
POMGNT1	NM_017739.3
POMGNT2	NM_032806.5
POMK	NM_032237.4
POMT1	NM_007171.3
POMT2	NM_013382.5
PREPL	NM_006036.4
PYGM	NM_005609.3
PYROXD1	NM_024854.3
RAPSN	NM_005055.4
RBCK1	NM_031229.3
RRM2B	NM_015713.4
RXYLT1	NM_014254.2

GENE	TRANSCRIPT
RYR1	NM_000540.2
SCN4A	NM_000334.4
SDHA*	NM_004168.3
SELENON	NM_020451.2
SGCA	NM_000023.2
SGCB	NM_000232.4
SGCD	NM_000337.5
SGCG	NM_000231.2
SIL1	NM_022464.4
SLC16A1	NM_003051.3
SLC18A3	NM_003055.2
SLC22A5	NM_003060.3
SLC25A20	NM_000387.5
SLC25A32	NM_030780.4
SLC5A7	NM_021815.2
SMCHD1	NM_015295.2
SMN1	NM_000344.3
SMN2	NM_017411.3
SNAP25	NM_130811.2
SPEG	NM_005876.4
SQSTM1	NM_003900.4
STAC3	NM_145064.2
STIM1	NM_003156.3
SUCLA2	NM_003850.2
SUCLG1	NM_003849.3
SUN1	NM_001130965.2
SUN2	NM_015374.2
SYNE1	NM_033071.3
SYNE2	NM_182914.2
SYT2	NM_177402.4
TANGO2	NM_152906.6
TAZ	NM_000116.4
TCAP	NM_003673.3
TIA1	NM_022173.2
TK2	NM_004614.4
TMEM43	NM_024334.2
TNNT1	NM_003283.5
TNNT3	NM_006757.3
TNPO3	NM_012470.3

GENE	TRANSCRIPT
TOR1AIP1	NM_001267578.1
TPM2	NM_003289.3
TPM3*	NM_152263.3
TRAPPC11	NM_021942.5
TRIM32	NM_012210.3
TSFM*	NM_001172696.1
TTN*	NM_001267550.2
TWNK	NM_021830.4
TYMP	NM_001953.4
VAMP1	NM_014231.3
VCP	NM_007126.3
VMA21	NM_001017980.3

Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with ≥50x depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated below. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. For some genes only targeted loci are analyzed (indicated in the table above). Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Confirmation of the presence and location of reportable variants is performed based on stringent criteria established by Invitae (1400 16th Street, San Francisco, CA 94103, #05D2040778), as needed, using one of several validated orthogonal approaches (PubMed ID 30610921). The following analyses are performed if relevant to the requisition. For PMS2 exons 12-15, the reference genome has been modified to force all sequence reads derived from PMS2 and the PMS2CL pseudogene to align to PMS2, and variant calling algorithms are modified to support an expectation of 4 alleles. If a rare SNP or indel variant is identified by this method, both PMS2 and the PMS2CL pseudogene are amplified by long-range PCR and the location of the variant is determined by Pacific Biosciences (PacBio) SMRT sequencing of the relevant exon in both long-range amplicons. If a CNV is identified, MLPA or MLPA-seq is run to confirm the variant. If confirmed, both PMS2 and PMS2CL are amplified by long-range PCR, and the identity of the fixed differences between PMS2 and PMS2CL are sequenced by PacBio from the long-range amplicon to disambiguate the location of the CNV. Technical component of confirmatory sequencing is performed by Invitae Corporation (1400 16th Street, San Francisco, CA 94103, #05D2040778). For C9orf72 repeat expansion testing, hexanucleotide repeat units are detected by repeat-primed PCR (RP-PCR) with fluorescently labeled primers followed by capillary electrophoresis. Interpretation Reference Ranges: Benign (Normal Range): <25 repeat units, Uncertain: 25-30 repeat units, Pathogenic (Full Mutation): >=31 repeat units. A second round of RP-PCR utilizing a non-overlapping set of primers is used to confirm the initial call in the case of suspected allele sizes of 22 or more repeats. Technical component of Fibroblast cell-culturing and gDNA extraction from skin punch biopsy is performed by Invitae Corporation (5 Technology Drive, Irvine CA 92618, #05D1052995).
- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at **http://www.ncbi.nlm.nih.gov/pubmed**.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC (**http://exac.broadinstitute.org**), gnomAD (**http://gnomad.broadinstitute.org**), and dbSNP (**http://ncbi.nlm.nih.gov/SNP**).
- A MedGen ID is a unique identifier referring to an article in MedGen, NCBI's centralized database of information about genetic disorders and phenotypes. Search by MedGen ID at **http://www.ncbi.nlm.nih.gov/medgen**. An OMIM number is a unique identifier referring to a comprehensive entry in Online Mendelian Inheritance of Man (OMIM). Search by OMIM number at **http://omim.org/**.
- Invitae uses information from individuals undergoing testing to inform variant interpretation. If "Invitae" is cited as a reference in the variant details this may refer to the individual in this requisition and/or historical internal observations.

Limitations

Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. Invitae's methods also detect insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. Invitae's deletion/duplication analysis determines copy number at a single exon resolution at virtually all targeted exons. However, in rare situations, single-exon copy number events may not be analyzed due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an

extracted genomic DNA sample. In very rare cases (such as circulating hematolymphoid neoplasm, bone marrow transplant, recent blood transfusion, or maternal cell contamination), the analyzed DNA may not represent the patient's constitutional genome.

CAPN3: Deletion/duplication analysis is not offered for exon 24. TSFM: Sequencing analysis is not offered for exon 5. FLNC: Deletion/duplication analysis is not offered for exon 47. Sensitivity and specificity for single nucleotide variants, insertions and deletions in exons 47-48 may be reduced due to the presence of segmental duplications overlapping the region. NEB: Deletion/duplication analysis is not offered for exons 82-105. NEB variants in this region with no evidence towards pathogenicity are not included in this report, but are available upon request. CTDP1: c.863+389C>T variant only. PGM1: Deletion/duplication analysis is not offered for exon 11. SDHA: Deletion/duplication analysis is not offered for this gene and sequencing analysis is not offered for exon 14. Sequencing analysis for exons 6-8 includes only cds +/- 10 bp. TTN: Exons 45-46, 147, 149, 164, 172-201 (NM_001267550.2) are excluded from analysis. TTN variants are included in the primary report based on functional effect and/or location. A complete list of variants of uncertain significance, likely benign and benign variants in TTN is available upon request. Variants are named relative to the NM_001267550.2 (meta) transcript. Variants in the coding sequence and intronic boundaries of the clinically relevant NM_133378.4 (N2A) and fetal isoforms are reported (PMID: 25589632, 29598826, 29691892, 31660661), with the exception of the PEVK tandem repeat region (172-198) (PMID: 28040389). SMN1 or SMN2: The SMN1 gene is identical to the SMN2 gene with the exception of exon 8 (typically referred to as exon 7). This assay unambiguously detects SMN1 exon 8 copy number and sequence variants. Sequence variants outside of exon 8 will also be detected, but this assay cannot determine whether the variant is located in SMN1 or SMN2. SMN2 exon 8 copy number will be reported for individuals with a positive result in SMN1. CNVs of exons 1-7 of SMN1 or SMN2 (typically referred to as exons 1-6 in the literature) will not be reported. Variants in all exons with no evidence towards pathogenicity are not reported, but are available upon request. This assay cannot detect silent carriers (individuals that have 2 functional copies of SMN1 on one chromosome and zero copies on the other). Therefore a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. For individuals with 2 copies of SMN1, the residual risk of being a carrier has been reported to be 1 in 121 in African Americans, 1 in 345 in Ashkenazi Jewish individuals, 1 in 628 in Asians, 1 in 632 in Caucasians, and 1 in 1061 in Hispanic individuals (PMID: 23788250). The SMA-STAT test does not detect sequence variants in SMN1 or SMN2, and therefore cannot be used to identify compound heterozygotes. TPM3: Deletion/duplication analysis is not offered for exon 10.

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Invitae. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Act (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

This report has been reviewed and approved by:

Thomas L. Winder, Ph.D., FACMG
Clinical Molecular Geneticist

INVITAE

VUS RESULTS GUIDE

What your results mean for you

No significant genetic changes ("pathogenic variants" or "mutations") were found in your genetic test. However, your test did find a genetic change called a variant of uncertain significance (VUS) in one or more of the genes tested. When we see a genetic change, but are unsure of its impact on health, it is called a variant of uncertain significance.

Right now, there is not enough information about the VUS to know whether it causes disease or not. A VUS is a common type of result. We all have many genetic changes that do not cause medical problems. Most of the time, we later learn that a VUS is not related to disease risk.

Your risk for disease could still be influenced by a combination of unidentified genetic, personal, lifestyle and/or environmental factors. So, it's important to talk to your healthcare provider if you have questions about your risk.

Create a plan with your healthcare provider

These genetic test results should be shared with your healthcare providers. The chance for you to develop a disease is not determined by genetic test results alone. Your provider can help you make informed decisions about your healthcare.

What your results mean for your family

Testing family members for a VUS is usually not recommended. However, your report will note if testing your family members will help us learn more about your specific VUS.

Although your genetic test did not find a significant genetic change, your family members have their own unique genetic makeup. Genetic testing can help them understand their overall chance of developing a genetic disease.

We (and others) are here to help

Genetic counseling can help you clearly and accurately understand your results so it's important to talk to your genetic counselor or other healthcare provider about your test results. Invitae also has board-certified genetic counselors who are available to answer questions about your test results or your personal or family medical history.

Log in to your patient portal (invitae.com) to view your results, search for a local or Invitae genetic counselor, or join Invitae's Patient Insight Network (PIN), a community where you can connect with other patients and share your experience.

This information in this results guide is meant to be used along with your genetic test results and other health information. It is not meant to replace a discussion with your healthcare provider and should not be considered or interpreted as medical advice.

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