INVITAE

detect
Muscular Dystrophy

ORDER ID
For Invitae internal use only

Requisition Form
Detect Muscular Dystrophy TRF956-2

This requisition form can be used to submit an order for **Detect Muscular Dystrophy**, a sponsored testing program for muscular dystrophy.

INSTRUCTIONS: Review the ordering options and then complete all sections of this form. Your ordering option will be indicated in the test selection section.

ORDERING OPTIONS

1. DETECT MUSCULAR DYSTROPHY PROGRAM

For individuals that meet the eligibility criteria below and wish to receive the program specific genetic testing panels.

REQUIRED: You must select below the appropriate eligibility criteria for this patient.

This program is available to individuals in the U.S. and Canada suspected of having a muscular dystrophy (must check at least one):

- ◯ Progressive muscle weakness
- ◯ Elevated CK levels
- ◯ Presumptive positive DMD from Newborn Screening Program
- ◯ Cardiac or respiratory involvement
- ◯ Calf hypertrophy or pseudohypertrophy
- ◯ Muscle biopsy showing dystrophic changes and/or immunohistochemical evidence for specific muscular dystrophy subtype
- ◯ Family history of muscular dystrophy*
  Clinical diagnosis, if known:

**Please note that this program is not intended for carrier screening of unaffected individuals.*

2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING

For relatives of program participants who received a Pathogenic/Likely Pathogenic result or approved VUS who want to receive gene specific family follow-up testing at no additional cost. Relatives do not need to meet the eligibility criteria listed above. Learn more at www.invitae.com/family.

PATIENT INFORMATION

First name	MI	Last name
Date of birth (MM/DD/YYYY)	Biological sex ◯ M ◯ F	MRN (medical record number)

Ancestry ◯ Asian ◯ Black/African American ◯ White/Caucasian ◯ Ashkenazi Jewish ◯ Hispanic ◯ Native American ◯ Pacific Islander ◯ French Canadian ◯ Sephardic Jewish ◯ Mediterranean ◯ Other: _____

Phone	Email address (report access after clinician releases)	
Address		City
State/Prov	ZIP/Postal code	Country

SPECIMEN INFORMATION

Specimen type: Blood (3-mL purple EDTA) -OR- Saliva (Oragene™) -OR- Assisted Saliva -OR- DNA source: _____

We are unable to accept blood/saliva from patients with:
- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Specimen collection date (MM/DD/YYYY): ☐☐ ☐☐ ☐☐☐☐
If not provided, the day before specimen receipt will be used

Special cases: ◯ History of/current hematologic malignancy in patient

INVITAE PARTNER CODE MDYS

CLINICIAN INFORMATION

Organization name		
Phone	Fax	
Address		City
State/Prov	ZIP/Postal code	Country

Primary clinical contact name (if different from ordering provider)	NPI
Primary clinical contact email address (for report access)	

Ordering provider (select one ordering provider by marking the checkbox before the name)

	Name	NPI	Email address (for report access)
◯			
◯			
◯			
◯			

Additional clinical or laboratory contacts (optional, to share access to order online)

◯ **Share this order with the primary clinical contact's default clinical team, manage at invitae.com**

Name	Email address (for report access)
Name	Email address (for report access)

CLINICAL HISTORY

FAMILY HISTORY

Is there a family history of disease for which the patient is being tested? ◯ Yes ◯ No If yes, describe below and attach pedigree and/or clinical notes.

Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis	Relative's relationship to this patient	Maternal or paternal	Diagnosed condition	Age at diagnosis

PERSONAL HISTORY

Is/was this patient affected or symptomatic?† ◯ Yes ◯ No

Provide details in the required clinical history questions (if applicable).

† Symptomatic means this patient has features or signs known or suspected to be related to the genetic testing being ordered and could include findings on physical examination, laboratory tests, or imaging.

OPTIONAL CLINICAL HISTORY (It is strongly encouraged to include notes, reports, and/or previous genetic test reults for this individual or affected family members. This information is useful for variant interpretation.)

MUSCULOSKELETAL	Y	N	UNKNOWN
Gait difficulties	◯	◯	◯
Ambulatory (if no, indicate age at which ambulation was lost below) Age at which ambulation was lost (if known): _____	◯	◯	◯
Scapular winging	◯	◯	◯
Calf hypertrophy	◯	◯	◯
Positive Gowers' maneuver	◯	◯	◯
Pain/cramps	◯	◯	◯
Joint contractures	◯	◯	◯
Asymmetry of weakness/wasting	◯	◯	◯
Foot drop	◯	◯	◯
Toe walking/achilles tendon tightness	◯	◯	◯
Inability to stand on toes	◯	◯	◯
Rippling muscle disease	◯	◯	◯
Diagnostic muscle biopsy (if yes, list specific findings below) Specific findings: _____	◯	◯	◯
RESPIRATORY			
Breathing difficulties	◯	◯	◯
Ventilator usage: day	◯	◯	◯
Ventilator usage: night	◯	◯	◯

CARDIAC	Y	N	UNKNOWN
Dilated cardiomyopathy	◯	◯	◯
Arrhythmia	◯	◯	◯
OCULAR			
Cataracts	◯	◯	◯
Severe myopia	◯	◯	◯
Retinal degeneration	◯	◯	◯
Ptosis or eye muscle weakness	◯	◯	◯
CENTRAL NERVOUS SYSTEM			
Learning disabilities	◯	◯	◯
Global developmental delays	◯	◯	◯
Motor delays only	◯	◯	◯
Nasal or dysarthric speech	◯	◯	◯
Intellectual disability	◯	◯	◯
SKIN FINDINGS			
Keratosis pilaris	◯	◯	◯
Abnormal scar formation (keloid or atrophic)	◯	◯	◯
Soft and velvety texture to the palmar skin of the hands and feet	◯	◯	◯
MEDICAL			
Corticosteroid Use	◯	◯	◯

OPTIONAL - REQUESTED VARIANTS FOR THIS PATIENT'S REPORT, IF KNOWN

*To have the presence or absence of specific variants commented on in this patient's report, provide the details below. For gene-specific family follow-up see **Note** under Test Selection.*

Was the proband (individual with variant) tested at Invitae? ◯ Yes, Invitae Order ID: RQ# _____ ◯ No: Attach copy of lab results (*required*)

Variant(s) *(e.g. GENE c.2200A>T (p.Thr734Ser) NM_00012345) If left blank, all variants identified in the proband will be commented on.*		**This patient's relationship to proband:**
		◯ Parent ◯ Sibling ◯ Grandchild
		◯ Child ◯ Self ◯ Other: _____

TEST SELECTION – Select option 1 or 2 below:

○ 1. DETECT MUSCULAR DYSTROPHY PROGRAM – Indicate one test below to be performed:

Test code	Test name		# of genes	Gene list
○ 03280	Invitae Comprehensive Neuromuscular Disorders Panel		109	ACTA1, AGRN, ALG2, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1S, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GFPT1, GMPPB, GNE, GYS1, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LDB3, LMNA, LMOD3, MATR3, MEGF10, MTM1, MUSK, MYH2, MYH7, MYL2, MYOT, MYPN, NEB, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, RAPSN, RXYLT1, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC5A7, SMN1, SMN2, SQSTM1, STAC3, STIM1, TAZ, TCAP, TIA1, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, VCP, VMA21
	○ 03280.1	Add-on preliminary-evidence genes	13	ALG14, HNRNPA2B1, HNRNPDL, LAMB2, LIMS2, LRP4, MYF6, SNAP25, SUN1, SUN2, SYNE1, SYNE2, TMEM43
	○ 03280.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1

Test code	Test name		# of genes	Gene list
○ 03291	Invitae Comprehensive Muscular Dystrophy Panel		48	ANO5, B3GALNT2, B4GAT,1 CAPN3, CAV3, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03291.1	Add-on preliminary-evidence genes	7	HNRNPDL, LIMS2, SUN1, SUN2, SYNE1, SYNE2, TMEM43
	○ 03291.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1

Test code	Test name		# of genes	Gene list
○ 03304	Invitae Limb-Girdle Muscular Dystrophy Panel		31	ANO5, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GAA, GMPPB, ISPD, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN
	○ 03304.1	Add-on preliminary-evidence genes	2	HNRNPDL, LIMS2
	○ 03304.2	Add-on facioscapulohumeral muscular dystrophy type 2 (FSHD2) gene	1	SMCHD1

Test code	Test name	# of genes	Gene list
○ 03301	Invitae Dystrophinopathies Test	1	DMD

○ 2. GENE-SPECIFIC FAMILY FOLLOW-UP TESTING *For relatives of a program participant ('proband') who received a Pathogenic/Likely Pathogenic result or approved VUS.*

Proband's Invitae Order ID:	This patient's relationship to proband:	Gene(s) to be tested in this patient:
RQ# _____	○ Parent ○ Sibling ○ Grandchild ○ Child ○ Other: _____	

***NOTE**: The presence or absence of all variants identified in the proband for the gene(s) ordered for gene-specific family follow-up will be commented on in this patient's report unless a limited selection is specified in the **Requested Variants** section above. Invitae will report any Pathogenic/Likely Pathogenic variants found in this patient for the gene(s) ordered.*

**Please note this program does not test for FSHD Type 1, OPMD, or Myotonic Dystrophy Types 1 and 2*
Invitae continually updates its panels based on the most recent evidence. If an order is placed using an outdated test requisition form, Invitae reserves the right to upgrade ordered tests to their current versions. Test IDs containing add-on codes will include the original panel as well as the add-on.

By signing this form, the medical professional acknowledges that the individual/family member authorized to make decisions for the individual (collectively, the "Patient") has been supplied information regarding and consented to undergo genetic testing, substantially as set forth in Invitae's Informed Consent for Genetic Testing (www.invitae.com/forms). In connection with the Program the Patient has been informed that Invitae may notify them of clinical updates related to genetic test results (in consultation with the ordering medical professional as indicated) and has been informed that de-identified Patient data may be used and shared with third parties, for research and commercial purposes and, in the U.S., to contact their medical professional. For orders originating in Canada, the Patient has been informed that their personal information and specimen will be transferred to and processed in the U.S. and that de-identified Patient data may be used and shared for research and commercial purposes in the U.S. The medical professional warrants that he/she will not seek reimbursement for this no-charge test from any third party, including but not limited to federal healthcare programs. The medical professional also hereby acknowledges that organization and clinician contact information provided in the order may be shared with third parties, including commercial organizations, that may contact the medical professional directly in connection with the Program, and that they have made the Patient aware that de-identified Patient data may be used and shared with such third parties, for purposes which include contacting their medical professional directly in connection with the Program. A list of third party partners may be provided upon request. I attest that I am authorized under applicable state law to order this test.

Medical professional signature (required)	Date (MM/DD/YYYY)