INVITAE detect

ORDER ID
For Invitae internal use only

Requisition Form

Invitae Detect LSDs Program TRF949-2

This requisition form can be used to submit a specimen for the Invitae Detect Lysosomal Storage Diseases (LSDs) program, a complimentary testing program for lysosomal storage disorders. Please confirm that the patient meets the eligibility requirements for the program. To submit orders for genetic testing outside of this program, please order through Invitae's online portal or use a standard requisition form, www.invitae.com/order-forms.

PROGRAM ELIGIBILITY:

This program is available to patients in the U.S. suspected of having a lysosomal storage disease (LSD) based on one or more of the following: clinical features, suspicion or known diagnosis of a specific LSD, family history of LSD, lab result suggestive of LSD, or presumptive positive NBS.

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

Address | City

State | Zip code | Country

SPECIMEN INFORMATION

Label each tube with the patient's full name, date of birth, and specimen collection date.
A requisition form MUST accompany each specimen. www.invitae.com/specimen-requirements

Specimen type : ◯ Blood ◯ Saliva ◯ Assisted saliva ◯ DNA - source:

DNA must be extracted in a CLIA or other suitably certified laboratory

We are unable to accept blood/saliva from patients with:

- *Allogeneic bone marrow transplants*
- *Blood transfusion <2 weeks prior to specimen collection*

Collection date (MM/DD/YYYY) *If not provided, date will be 1 day prior to our receipt of specimen. For DNA, provide date retrieved from archive.*

Special cases : ◯ History of/current hematologic malignancy

REASON FOR TESTING

Previous results *(if applicable and not included in clinical criteria- enclose copy of report)*

ORGANIZATION INFORMATION

Organization name and address

Organization name

Phone | Fax

Address | City

State | Zip code | Country **United States**

Primary clinical contact

Name | Role/title

Phone | NPI

Email address (for report access)

Ordering physician

◯ **Same as primary clinical contact**

Name | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

INVITAE PARTNER CODE LYSO

INVITAE DETECT LSDs PROGRAM ELIGIBILITY/CLINICAL INFORMATION

Suspicion of lysosomal storage disorder based on (check any and all that apply):

☐ **Clinical features (see symptom list on page 2)**

☐ **Suspicion or known diagnosis of a specific lysosomal storage disease (specify disorder on page 2)**

Age at onset: _____ Clinical diagnosis: _____

☐ **Family history of lysosomal storage disease**

Specify disorder: _____

☐ **Lab result suggestive of lysosomal storage disease**

☐ Elevated GAGs: Specify type(s) _____

☐ Abnormal LSD enzyme analysis: Enzyme: _____ Patient value: _____ Reference range: _____

☐ **Presumptive positive NBS disorder**

Specify disorder: _____

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INVITAE DETECT LSDs PROGRAM ELIGIBILITY/CLINICAL INFORMATION (continued)

If you selected "clinical features" on page 1, select all that apply in the following list:

Heart:
- ☐ Evidence of storage on heart biopsy
- ☐ Dilated cardiomyopathy
- ☐ Hypertrophic cardiomyopathy
- ☐ Valvular disease

Gastrointestinal:
- ☐ Evidence of storage on liver biopsy
- ☐ Hepatosplenomegaly/ Hepatomegaly/ Splenomegaly

Kidney:
- ☐ Chronic kidney failure
- ☐ Evidence of storage on kidney biopsy
- ☐ Proteinuria of unknown etiology

Nervous system:
- ☐ Acroparesthesisa
- ☐ Macrocephaly
- ☐ Limb girdle muscular dystrophy
- ☐ Regression of milestones
- ☐ Seizures
- ☐ Stroke

List other relevant clinical information (symptoms, imaging studies, etc):

_____

_____

_____

_____

Eye:
- ☐ Cherry red spot
- ☐ Corneal clouding
- ☐ Corneal verticillata
- ☐ Horizontal gaze palsy
- ☐ Oculomotor apraxia
- ☐ Retinal blindness
- ☐ Retinal/scleral vessel tortuosity
- ☐ Supranuclear gaze palsy

Skeletal:
- ☐ Bone crisis
- ☐ Carpal tunnel in childhood
- ☐ Dysostosis multiplex
- ☐ Erlenmeyer flask deformity
- ☐ Focal lytic/sclerotic lesions
- ☐ Gibbus deformity
- ☐ Pathological fractures not related to cancer
- ☐ Short stature

Other:
- ☐ Angioketatomas
- ☐ Anhydrosis/hypohydrosis
- ☐ Facial coarsening
- ☐ Hirsutism
- ☐ Hearing loss (conductive, sensorineural, or mixed)
- ☐ Unexplained tinnitus

If you selected "suspicion or known diagnosis of a specific lysosomal storage disease" on page 1, specify disorder in the following list:

- ☐ alpha-mannosidosis
- ☐ Aspartylglucosaminuria
- ☐ beta-mannosidosis
- ☐ cystinosis
- ☐ Danon disease
- ☐ Fabry disease
- ☐ Farber disease
- ☐ Fucosidosis
- ☐ Galactosialidosis
- ☐ GM1 gangliosidosis, Mucopolysaccharidosis IVb
- ☐ GM2-gangliosidosis, AB variant
- ☐ Infantile sialic acid storage disease, Salla disease
- ☐ Krabbe disease
- ☐ Lysosomal acid lipase deficiency
- ☐ Metachromatic leukodystrophy
- ☐ Mucolipidosis III gamma
- ☐ Mucolipidosis type I, Sialidosis I
- ☐ Mucolipidosis type II alpha/beta, Mucolipidosis III alpha/beta
- ☐ Mucolipidosis type IV
- ☐ Mucopolysaccharidosis IIIA
- ☐ Mucopolysaccharidosis IIIB
- ☐ Mucopolysaccharidosis type I
- ☐ Mucopolysaccharidosis type II
- ☐ Mucopolysaccharidosis type IIIC
- ☐ Mucopolysaccharidosis type IIID
- ☐ Mucopolysaccharidosis type IVa
- ☐ Mucopolysaccharidosis type IX
- ☐ Mucopolysaccharidosis type VI
- ☐ Mucopolysaccharidosis type VII
- ☐ Multiple sulfatase deficiency
- ☐ Neuronal ceroid lipofuscinosis 1 (CLN1)
- ☐ Neuronal ceroid lipofuscinosis 10 (CLN10)
- ☐ Neuronal ceroid lipofuscinosis 14 (CLN14)
- ☐ Neuronal ceroid lipofuscinosis 2 (CLN2)
- ☐ Neuronal ceroid lipofuscinosis 3 (CLN3)
- ☐ Neuronal ceroid lipofuscinosis 5 (CLN5)
- ☐ Neuronal ceroid lipofuscinosis 6 (CLN6)
- ☐ Neuronal ceroid lipofuscinosis 7 (CLN7)
- ☐ Neuronal ceroid lipofuscinosis 8 (CLN8)
- ☐ Niemann Pick types A and B
- ☐ Niemann-Pick type C
- ☐ Pompe disease
- ☐ Prosaposin deficiency, SapA deficiency (Krabbe variant), SapB deficiency (MLD variant), SapC deficiency (Gaucher variant)
- ☐ Pycnodysostosis
- ☐ Sandhoff disease
- ☐ Schindler disease
- ☐ Tay-Sachs disease

ORDER INSTRUCTIONS

All tests on this form are organized by clinical area. If your order contains tests from multiple clinical areas, you will need to send in two sample tubes and your order will represent two billable events. Your test results will be delivered as two reports. Please contact Client Services with any questions. For Invitae's full test menu, please visit **www.invitae.com**.

Invitae continually updates its panels based on the most recent evidence. Please note that if an order is placed using an older version of this form, Invitae reserves the right to upgrade any ordered panel(s) to the current version(s).

CLINICAL AREA: METABOLIC AND IMMUNOLOGY

Test code	Test name	# gene(s)	Gene list
◯ 06170	Invitae Comprehensive Lysosomal Storage Disorders Panel*	48	AGA, ARSA, ARSB, ASAH1, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSK, FUCA1, GAA, GALC, GALNS, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1
	◯ 06170.1 Add-on chitotriosidase deficiency gene	1	CHIT1
	◯ 06170.2 Add-on preliminary-evidence gene	1	ATP13A2
	◯ 06170.3 Add-on adult-onset neuronal ceroid lipofuscinoses genes	3	CTSF, DNAJC5, GRN
◯ 06185	Invitae Comprehensive Mucopolysaccharidoses (MPS) Panel	11	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH
	◯ 06185.1 Add-on mucolipidosis and oligosaccharidoses genes	12	AGA, CTSA, CTSK, FUCA1, GNPTAB, GNPTG, MAN2B1, MANBA, MCOLN1, NAGA, NEU1, SLC17A5
◯ 06183	Invitae Wilson Disease Test	1	ATP7B

**This panel does not currently test for Gaucher disease.*

▶ INDIVIDUAL GENES

◯ AGA	◯ ARSA	◯ ARSB	◯ ASAH1	◯ CLN2 (TPP1)	◯ CLN3	◯ CLN5	◯ CLN6
◯ CLN8	◯ CTNS	◯ CTSA	◯ CTSD	◯ CTSK	◯ FUCA1	◯ GAA	◯ GALC
◯ GALNS	◯ GLA	◯ GLB1	◯ GM2A	◯ GNPTAB	◯ GNPTG	◯ GNS	◯ GUSB
◯ HEXA	◯ HEXB	◯ HGSNAT	◯ HYAL1	◯ IDS	◯ IDUA	◯ KCTD7	◯ LAMP2
◯ LIPA	◯ MAN2B1	◯ MANBA	◯ MCOLN1	◯ MFSD8	◯ NAGA	◯ NAGLU	◯ NEU1
◯ NPC1	◯ NPC2	◯ PPT1	◯ PSAP	◯ SGSH	◯ SLC17A5	◯ SMPD1	◯ SUMF1

CLINICAL AREA: CARDIOLOGY/NEUROLOGY

Test code	Test name	# gene(s)	Gene list
◯ 02251	Invitae Cardiomyopathy Comprehensive Panel	50	ABCC9, ACTC1, ACTN2, AGL, BAG3, CACNA1C, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL
	◯ 02251.1 Add-on preliminary-evidence genes	31	ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, ILK, JPH2, LAMA4, LDB3, LRRC10, MED12, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TGFB3, TMPO, TXNRD2
	◯ 02251.2 Add-on RASopathy genes not included in panel	17	A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1
	◯ 02251.3 Add-on autosomal recessive syndromic pediatric cardiomyopathy genes	8	ACADVL, ALMS1, CPT2, DNAJC19, ELAC2, MTO1, SDHA, TMEM70

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CLINICAL AREA: CARDIOLOGY/NEUROLOGY

Test code	Test name		# gene(s)	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

RE-REQUISITION

Invitae offers one re-requisition at no additional charge within 90 days for genes related to the original clinical area. For more information and to request online, please visit **www.invitae.com/re-requisition**.

ASSAY

Invitae is a CAP-accredited and CLIA-certified clinical diagnostic laboratory performing full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS). Search for details on the analysis of any gene in our test catalog at **www.invitae.com/physician/search**.

To request a complimentary specimen collection kit, visit **www.invitae.com/request-a-kit.**

SHIPPING INSTRUCTIONS

Please ship specimen to Invitae:

Attn: Invitae Client Services
1400 16th Street
San Francisco, CA 94103 USA

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/patient-consent) and in connection with the Program, and 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). 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. In addition to the above, I attest that I am the ordering physician, or I am authorized by the ordering physician to order this test, or I am authorized under applicable state law to order this test.

Medical professional signature (required)	Date