Invitae Detect Lysosomal Storage Diseases (LSDs) Program Eligibility Criteria

FM189-2

Patients in the U.S. with a suspected or confirmed diagnosis of a lysosomal storage disease (LSD) are invited to take part in Invitae Detect LSDs, a sponsored genetic testing program brought to you by Invitae Corporation.

The patient must meet the eligibility criteria below to qualify for the program. Please fill out, print, and sign this form and include it when sending in the specimen. Additional information about the ordering process can be found at www.invitae.com/detectLSDs.

To submit orders for genetic testing outside of this program, please order through Invitae's online portal or use a standard requisition form, accessible at www.invitae.com/order-forms.

Patient name	Date of birth (MM/DD/YYYY)

INVITAE DETECT LSDs PROGRAM ELIGIBILITY CRITERIA

This program is available to patients in the U.S. suspected of having a lysosomal storage disease based on one or more of the following:

☐ **Clinical features (see symptom list below)**

☐ **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: _____

If you selected "clinical features" above, 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

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INVITAE DETECT LSDs PROGRAM ELIGIBILITY/CLINICAL INFORMATION (continued)

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

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. 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**