

detect

**FREE
MLD sequencing**



Invitae Detect Lysosomal Storage Diseases (LSDs) Genetic Testing Program

SPONSORED, NO-CHARGE GENETIC TESTING AND COUNSELING
FOR PATIENTS SUSPECTED OF HAVING AN LSD



The Invitae Detect LSDs program offers sponsored no-charge genetic testing and counseling to diagnose LSDs, bringing patients closer to an accurate diagnosis and appropriate clinical management.



Influence clinical outcomes

Diagnose LSDs faster to enable improved clinical outcomes.



Impact quality of life

Get patients the right information quicker to regain valuable treatment time.



Break down barriers to genetic testing

Both testing and counseling for this program are sponsored, no-charge offerings.



Give patients more than just test results

If the test is positive, the proband's blood relatives are eligible for the program using program code LYSO.

WHAT ARE LSDs?

- Progressive, multisystemic inherited metabolic diseases
- Associated with high morbidity and mortality
- Approximately 50 known LSDs

PROGRAM ELIGIBILITY

To qualify for no-charge testing through the Invitae Detect LSDs program, the individual must be suspected of having an LSD based on at least one of the following:

- Clinical features
- Suspicion of, or known diagnosis of, a specific LSD
- Family history related to LSDs
- Lab result suggestive of LSDs
- Presumptive positive newborn screening

GENETIC TESTING WITH INVITAE

Invitae offers LSDs testing with multiple panels as well as with single genes:

- Comprehensive Lysosomal Storage Disorders Panel*
- Comprehensive Mucopolysaccharidoses (MPS) Panel
- Or test the following single genes:
- Comprehensive Neuromuscular Disorders Panel
- Cardiomyopathy Comprehensive Panel

THE ARSA GENE CAUSES MLD

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

You can also order the Invitae Canavan Disease Test and the Invitae Wilson Disease Test.

*This panel does not currently test for Gaucher disease.

GENETIC COUNSELING

All patients enrolled in the Invitae Detect LSDs program will be eligible for sponsored, no-charge post-test genetic counseling with GeneMatters, a third-party genetic counseling service. For assistance with genetic counseling or to schedule an appointment, please contact GeneMatters at 1-866-741-5331, or online at www.genematters.com (use partner code: LYSO).

For more information and access to a portal where your doctor can order MLD sequencing, please visit [invitae.com/detectLSDs](https://www.invitae.com/detectLSDs).

While third parties and commercial organizations may provide financial support for this program, tests and services are performed by Invitae. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing and counseling are available in the US and Canada. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any other products or services from Invitae or from third parties or commercial organizations.