

# 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 outcomes

Diagnose LSDs faster to enable optimal patient management.



## 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 **70 known LSDs**

## PROGRAM ELIGIBILITY

To qualify for no-charge testing through the Invitae Detect LSDs program, the individual must be located in the US or Canada and 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

## References

- Meikle PJ and Grasby DJ. *Mol Genet Metab.* 88(4):307-14 (2006)
- Platt FM et al. *Nat Rev Dis Primers.* 4(1):27 (2018)

## 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

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 Wilson Disease Test.

## GENETIC COUNSELING\*

Individuals in the US & Canada tested through the Detect LSDs program are eligible for post-test genetic counseling through Invitae at no charge as part of the program. Patients can contact Invitae Client Services at 1-800-436-3037 and ask to schedule a genetic counseling appointment.

**For more information or to order a test, 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 (pseudonymized) 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 is available in Canada and the US only. 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.*