

# SUSPECT GAUCHER DISEASE?

# SUSPECT ASMD?

historically known as Niemann-Pick disease types A, A/B, and B

**Gaucher disease and ASMD are rare, progressive, genetic diseases with significant phenotypic overlap<sup>1,2</sup>**

Frequency of key symptoms in patients with

**Gaucher disease type 1:**

**ASMD types A/B and B<sup>4</sup>:**

**87%**



**SPLENOMEGALY**

**>90%**

**79%**



**HEPATOMEGALY**

**>70%**

**56%**



**THROMBOCYTOPENIA**

**>50%**

## Gaucher disease and ASMD: causes and impact on patients

### Gaucher disease:

Lysosomal accumulation of glucocerebroside (GL-1) in cells of monocyte-macrophage lineage.<sup>5</sup>

### Underlying cause

### ASMD:

Deficiency of the enzyme acid sphingomyelinase (ASM), resulting in the lysosomal accumulation of sphingomyelin in the monocyte-macrophage system.<sup>4</sup>

Panethnic, affecting both children and adults. Gaucher disease type 1 has a prevalence of 1:40,000 world-wide but is **~1:850 in patients of Ashkenazi Jewish ancestry**.<sup>1,5</sup>

### Patient population

Panethnic, affecting both children and adults. Among patients suspected of having Gaucher, an ASMD diagnosis is not uncommon.<sup>4,6</sup>

Symptoms affect major organs and may significantly impact quality of life and **can lead to a shortened lifespan**.<sup>1</sup>

### Disease burden

By the age of 35, ASMD type A/B and B patients have **~30% reduced survival probability** compared to the US general population.<sup>7\*</sup>

## PARALLEL TEST TO KNOW.

**Expert guidelines recommend parallel testing for Gaucher disease and ASMD<sup>2</sup>**

See next page for more information on testing and diagnosis.

\*Data extrapolated from a Kaplan-Meier curve generated in an 11-year natural history study that evaluated morbidity and mortality in 59 patients with ASMD type A/B and B. At entry, 30 patients were in the pediatric age group (<18 years of age) and 29 patients were adults (≥18 years of age). There were 9 deaths during the follow-up period. Reduction in survival probability is absolute, not relative. US general population as of 2017.

# PARALLEL TEST TO KNOW

# DIAGNOSTIC TESTING IS SIMPLE

Some laboratories offering Gaucher and ASMD testing are listed below. There may be other laboratory tests appropriate for your patient and this is not an endorsement of any one lab. Other testing options can be found at [www.concertgenetics.com](http://www.concertgenetics.com) or [www.ncbi.nlm.nih.gov/gtr](http://www.ncbi.nlm.nih.gov/gtr). Consult each laboratory for a full range of options. Content is current at time of printing and tests may not be available in all states; please contact the specific laboratory to confirm test availability and all logistics. Sanofi does not review or control the content of non-Sanofi websites. These listings do not constitute an endorsement by Sanofi of information provided by any other organizations.

Lab	Available Testing	GD	ASMD	Sample Requirements	Kits	Avg TAT	Blood Draw	Billing	Contact
Centogene	Enzyme	✓	✓	WB: 5ml EDTA (lavender) tube; DBS card: 10 circles	Blood, DBS, Saliva	7 d	Yes	Inst, Self-pay, Ins	P: 617-580-2102 E: <a href="mailto:customer.support-US@centogene.com">customer.support-US@centogene.com</a> W: <a href="http://www.centogene.com">www.centogene.com</a>
	Sequencing (+/- Del/Dup)	✓	✓	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab		15 d			
	Lyso-GL-1	✓	✓	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles		7 d			
	LSM-509	✓	✓	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles		7 d			
Greenwood Genetic Center	Enzyme	✓	✓	WB: 5-10 ml (3-5 ml for ASMD) sodium heparin (green) tube; DBS card: 3 circles	Blood, DBS, Saliva	2 wks	No	Inst, Self-pay, Ins (SC residents)	P: 800-473-9411 E: <a href="mailto:labgc@ggc.org">labgc@ggc.org</a> W: <a href="http://www.ggc.org">www.ggc.org</a>
	Sequencing	✓	✓	WB: 5-6 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva		3 wks			
Labcorp	Enzyme	✓	✓	WB: 2x 10ml EDTA (lavender) tube (peds 1 x 10ml) Note: Labcorp Test Code: 451780; Integrated Test Code: 370	Blood, Buccal	3-13 d	Yes	Inst Self-Pay, Ins	Labcorp Customers: P: 800-345-4363 W: <a href="http://www.labcorp.com">www.labcorp.com</a>
	Sequencing	✓	✓	WB: 7 ml EDTA (lavender) or ACD (yellow) tube Note: For sequencing done via "Inheritest Gene-Specific Sequencing" (test code: 451910). Indicate GBA or SMPD1 gene as needed.		9-15 d			
The Lantern Project (PerkinElmer Genomics)	Enzyme	✓	✓	WB: 2-10 ml sodium heparin (green) tube (volume varies with age); DBS card: 3 circles	Blood, DBS, Saliva	3 d	Yes	No charge*	P: 866-354-2910 E: <a href="mailto:genomics@perkinelmer.com">genomics@perkinelmer.com</a> W: <a href="http://www.LanternProjectDx.com">www.LanternProjectDx.com</a>
	Sequencing	✓	✓	WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles; Saliva		3 wks			
	Lyso-GL-1	✓	✓	WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles		3 d			
Mayo Clinic Laboratories	Enzyme	✓	✓	WB: 6 ml ACD (yellow) tube	DBS, Saliva	5-10 d	Yes	Inst, Ins (can be billed in some cases but account required)	P: 800-533-1710 E: <a href="mailto:mcl@mayo.edu">mcl@mayo.edu</a> W: <a href="http://www.mayocliniclabs.com">www.mayocliniclabs.com</a>
	Sequencing	✓	✓	WB: 3 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2-5 circles		14-20 d			
	Lyso-GL-1	✓	✓	WB: 1 ml EDTA (lavender), ACD B (yellow) or sodium heparin (green); Plasma: 0.3 ml; DBS card: 2 circles (Note: Order codes GPSY, GPSYP, or GPSYW)		2-8 d			
	Oxysterols	✓	✓	WB: 1ml EDTA (lavender), ACD B (yellow) or sodium heparin (sodium or lithium) (green) tubes; Frozen plasma: min 0.25 ml; DBS card: 2 circles		2-8 d			

\*Testing is performed at no charge. Local charges may apply for sample collection, processing, or shipping.

ACD=acid citrate dextrose; Avg TAT=average turnaround time; d=days; DBS=dried blood spot; Del=deletion; Dup=duplication; EDTA=ethylenediaminetetraacetic acid; Ins=insurance; Inst=institution; LSM=lyso-sphingomyelin; WB=whole blood; wks=weeks.

**References:** 1. Mistry PK, Cappellini MD, Lukina E, et al. A reappraisal of Gaucher disease-diagnosis and disease management algorithms. *Am J Hematol*. 2011;86(1):110-115. 2. McGovern MM, Dionisi-Vici C, Giugliani R, et al. Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. *Genet Med*. 2017;19(9):967-974. 3. Charrow J, Andersson HC, Kaplan P, et al. The Gaucher Registry: demographics and disease characteristics of 1698 patients with Gaucher disease. *Arch Intern Med*. 2000;160(18):2835-2843. 4. McGovern MM, Avetisyan R, Sanson BJ, Lidove O. Disease manifestations and burden of illness in patients with acid sphingomyelinase deficiency (ASMD). *Orphanet J Rare Dis*. 2017;12(1):41. 5. Grabowski GA, Petsko GA, Kolodny EH. Chapter 146: Gaucher disease. In: Valle D, Beaudet AL, Vogelstein B, eds. *The Online Metabolic and Molecular Bases of Inherited Disease*. New York, NY: McGraw Hill; 2014. <http://ommbid.mhmedical.com/content.aspx?bookid=474&sectionid=45374148>. Accessed March 2022. 6. Lukacs Z, Cobos PN, Murko S, et al. Multiplexed testing for Gaucher, Niemann Pick A/B disease and acid lipase deficiency. Presented at: 14th Annual WorldSymposium™; February 5-9, 2018; San Diego, California. 7. McGovern MM, Wasserstein MP, Bembi B, et al. *Orphanet J Rare Dis*. 2021; 16 (212):1-14.



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