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[1,2]

Frequency of key symptoms in patients with

Gaucher disease[1,3]:		ASMD type B[4]:
87%	 SPLENOMEGALY	>90%
79%	 HEPATOMEGALY	>70%
56%	 THROMBOCYTOPENIA	>50%

Gaucher disease and ASMD: causes and impact on patients

Gaucher disease type 1:		ASMD:
Lysosomal accumulation of glucocerebroside (GL-1) in cells of monocyte-macrophage lineage.[5]	**Underlying cause**	Deficiency of the enzyme acid sphingomyelinase (ASM), resulting in the lysosomal accumulation of sphingomyelin in the monocyte-macrophage system.[4]
Panethnic (frequency of ~1:40,000) but with a frequency of **~1:850 in patients of Ashkenazi Jewish ancestry**. Affects both children and adults.[1,5]	**Patient population**	Panethnic, affecting both children and adults. Among patients suspected of having Gaucher, an ASMD diagnosis is not uncommon.[4,6]
Symptoms affect major organs and may significantly impact quality of life and **can lead to a shortened lifespan**.[1]	**Disease burden**	By the age of 35, ASMD type B patients have **~30% reduced survival probability** compared to the US general population.[7]*

PARALLEL TEST TO KNOW.

Expert guidelines recommend parallel testing for Gaucher disease and ASMD[2]

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 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 of the laboratories offering diagnostic testing for ASMD and Gaucher disease are listed below. There may be other diagnostic testing appropriate for your patient, and this is not an endorsement of any specific lab. Other testing options can be found at www.concertgenetics.com or www.ncbi.nlm.nih.gov/gtr. Consult each laboratory for a full range of options. Content is current at time of publication, and tests may not be available in all states; please call the laboratory to confirm test availability, sample shipping information, and all other logistics. Sanofi does not review or control the content of non–Sanofi websites. This listing does 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:** customer.support-US@centogene.com **W:** www.centogene.com
	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:** labgc@ggc.org **W:** www.ggc.org
	Sequencing	√	√	WB: 5-6 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva		3 wks			
LabCorp/ Integrated Genetics	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:** www.labcorp.com **Integrated Customers:** **P:** 800-848-4436 **E:** asklGclientservices@integratedgenetics.com **W:** www.integratedgenetics.com
	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			
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:** mcl@mayo.edu **W:** www.mayocliniclabs.com
	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			
Sema4	Enzyme	√	√	WB: 5-10 ml sodium heparin (green) tube	Blood, Saliva	7 d	Yes	Inst, Self-Pay, Ins	**P:** 800-298-6470 **E:** clientservices@sema4.com **W:** www.sema4.com
	Sequencing	√	√	WB: 2 x 5-10 ml ACD (yellow) OR 2 x 5-10 ml EDTA (lavender); Saliva		14 d			
	Lyso-GL-1	√		WB: 1-2 ml EDTA (lavender) or heparin (green) tube; Frozen plasma: 0.5-1 ml		5 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 World*Symposium*™; 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.