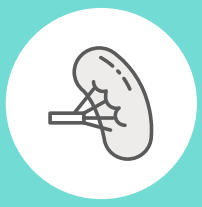
SUSPECT ASMD? TEST TO KNOW.

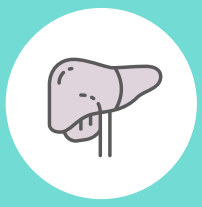
ASMD (acid sphingomyelinase deficiency), historically known as **Niemann-Pick disease types A, A/B, and B,** is a **progressive and potentially life-threatening genetic disease** that affects both children and adults.[1]

If you see a combination of these signs and symptoms, include ASMD in your differential[1,2]:



Splenomegaly

Interstitial lung disease



Hepatomegaly

Thrombocytopenia

Gastrointestinal issues

Pediatric growth delay

- ASMD is a lysosomal storage disease caused by **deficiency of the enzyme acid sphingomyelinase (ASM)**, resulting in the buildup of the substrate sphingomyelin[1]
- Accumulation of sphingomyelin in major organs can lead to **progressive, multisystemic damage and early death[1]**

By the age of 35, ASMD type B patients have **~30%-reduced survival probability** compared to the US general population.[3]*

DIAGNOSTIC TESTING FOR ASMD IS SIMPLE.[4]

Perform an ASM biochemical enzyme assay on isolated peripheral blood leukocytes, dried blood spots (DBS), or skin fibroblasts†

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Low residual ASM activity

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ASMD diagnosis confirmed

Additional diagnostic confirmation can be achieved using molecular genetic testing

Adapted from McGovern MM et al. *Genet Med.* 2017;19(9):967-974.[4]

You can play a critical role in the early diagnosis of ASMD, enabling appropriate symptom management and family screening.[1,4]

Visit **ASMDfacts.com/hcp** for more information on diagnosis and testing

Please see next page for list of labs where ASM enzyme tests and DNA tests can be performed ▶

*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.

†Limitations to DBS testing include the potential effects of anemia and recent transfusions on results. Skin fibroblasts or *SMPD1* gene sequencing can be used in equivocal cases.

TESTING OPTIONS FOR ASMD

Some of the laboratories offering diagnostic testing for ASMD 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	SAMPLE REQUIREMENTS	KITS	AVG TAT	MOBILE BLOOD DRAW	BILLING	CONTACT
Centogene	Enzyme	WB: 5 mL 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: 1 mL EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab		15 d			
	LSM-509	WB: 1 mL EDTA (lavender) tube; DBS card: 10 circles		7 d			
LabCorp/ Integrated Genetics	Enzyme	WB: 2x 10 mL EDTA (lavender) tube (peds 1 x 10 mL). 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:** askIGclientservices@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			
Greenwood Genetic Center	Enzyme	WB: 3-5 mL sodium heparin (green) tube; DBS card: 3 circles	Blood, DBS, Saliva	2 wks	No	Inst, Self-pay, Ins (South Carolina residents only)	**P:** 800-473-9411 **E:** labgc@ggc.org **W:** www.ggc.org
	Sequencing	WB: 5-7 mL EDTA (lavender) tube; DBS card: 3 circles; Saliva		3 wks			
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 is 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			
	Oxysterols	WB: 1 mL EDTA (lavender), ACD B (yellow) or sodium heparin (sodium or lithium) (green) tubes; Frozen plasma: 0.25 mL; DBS card: 2 circles		2-8 d			
Sema4	Enzyme	WB: ACD (yellow) preferred, sodium heparin or EDTA accepted	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			

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

References: 1. 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. **2.** Cox GF, Clarke LA, Giugliani R. *JIMD Rep*. 2018;41:119-129. **3.** McGovern MM Wasserstein MP, Bembi B, et al. *Orphanet J Rare Dis*. 2021; 16 (212):1-14. **4.** 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.

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