ASMD TESTING
Find your diagnosis

ASMD
ACID SPHINGOMYELINASE DEFICIENCY

Experiencing symptoms of ASMD? Ask your doctor about getting tested

ASMD (acid sphingomyelinase deficiency)—also known as Niemann-Pick disease types A, A/B, and B—is a rare, inherited disease. The symptoms of ASMD can affect individuals differently and may worsen over time, potentially leading to serious health issues.

The most common symptoms you may see or feel:

- Protruding abdomen
- Coughing and difficulty breathing
- Easy bruising and bleeding
- Abdominal pain and diarrhea

Other symptoms you may experience include:

- Fatigue
- Higher than normal liver enzyme levels
- Abnormal cholesterol levels
- Growth delay and/or developmental delays in children

Symptoms of ASMD can vary among patients, affecting some more than others.

Family member diagnosed with ASMD? Ask your doctor about getting tested

ASMD is an inherited condition that parents can pass on to their children—even if the parents don't have ASMD. Because ASMD runs in families, if one member of a family has the disease, others may too.

Family screening for ASMD can help get to a diagnosis quicker so that symptoms can be managed earlier.

Getting tested for ASMD is simple, and an early diagnosis can be the first step to appropriately managing your symptoms.

ANNE
Living with ASMD

PLEASE SEE OTHER SIDE FOR LIST OF LABS WHERE ASMD DIAGNOSTIC TESTS CAN BE PERFORMED ▶

Testing information for you and your doctor

Getting tested for ASMD is simple

- ASMD is a lysosomal storage disorder caused by a deficiency of the ASM enzyme
- A blood test that measures the amount of ASM enzyme in your blood can confirm an ASMD diagnosis
  - ASMD biomarker tests can measure other indicators of ASMD in your blood
- Additional confirmation may be done using genetic testing (either a blood or saliva test)
- Invasive testing procedures, such as bone marrow biopsies, are NOT required to confirm a diagnosis of ASMD

Some of the laboratories offering diagnostic testing for ASMD are listed below.

Lab	Available testing	Average time to results	Contact
Centogene	ASM Enzyme Activity	7 days	P: 617-580-2102 E: customer.support-US@centogene.com W: centogene.com
	Genetic Testing	15 days	
	ASMD Biomarker	7 days	
EGL Genetics/ Eurofins	Genetic Testing	2-6 weeks	P: 855-831-7447 E: eglcs@egl-eurofins.com W: egl-eurofins.com
Greenwood Genetic Center	ASM Enzyme Activity	2 weeks	P: 800-473-9411 E: labgc@ggc.org W: ggc.org
	Genetic Testing	3 weeks	
LabCorp/ Integrated Genetics	Genetic Testing	9-15 days	LabCorp Customers: P: 800-345-4363 W: labcorp.com Integrated Customers: P: 800-848-4436 E: asklGclientservices@integratedgenetics.com W: integratedgenetics.com
Mayo Clinic Laboratories	Genetic Testing	14-20 days	P: 800-533-1710 E: mcl@mayo.edu W: mayocliniclabs.com
	ASMD Biomarker	2-8 days	
National Gaucher Foundation	ASM Enzyme Activity	Not applicable	W: gaucherdisease.org/intakeform
	Genetic Testing		
Seattle Children's Hospital	Genetic Testing	2-3 weeks	P: 206-987-2617 E: labGC@seattlechildrens.org W: seattlechildrenslab.testcatalog.org
Sema4	ASM Enzyme Activity	7 days	P: 800-298-6470 E: clientservices@sema4.com W: sema4.com
	Genetic Testing	14 days	

There may be other appropriate diagnostic tests and this is not an endorsement of any specific lab. Other testing options can be found at concertgenetics.com or 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 Genzyme does not review or control the content of non–Sanofi Genzyme websites. This listing does not constitute an endorsement by Sanofi Genzyme of information provided by any other organizations.

Suspect ASMD? Get tested

If you are experiencing any symptoms of ASMD or a family member has been diagnosed, talk to your doctor about testing.

For more information about ASMD, visit ASMDfacts.com or please reach out to us at 1-800-745-4447, Option 3 for Patient Services, then press 1 for more information on ASMD

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