

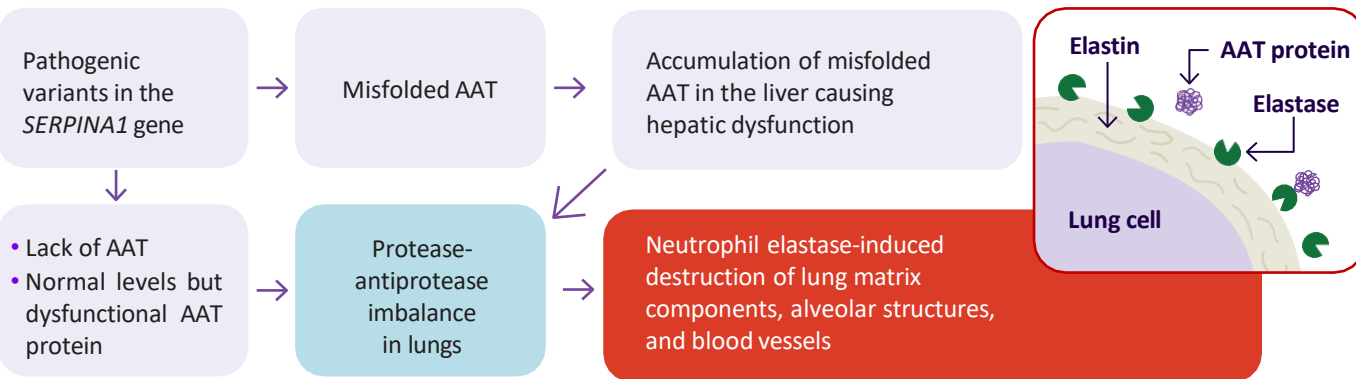


Alpha-1 antitrypsin deficiency (AATD): An underdiagnosed cause of chronic obstructive pulmonary disease (COPD)

Pulmonary Manifestations of AATD

AATD is an underdiagnosed genetic disease. Severe disease is characterized by accelerated pulmonary deterioration and/or liver dysfunction^{1,2}

What is the etiology of AATD?^{1,2}

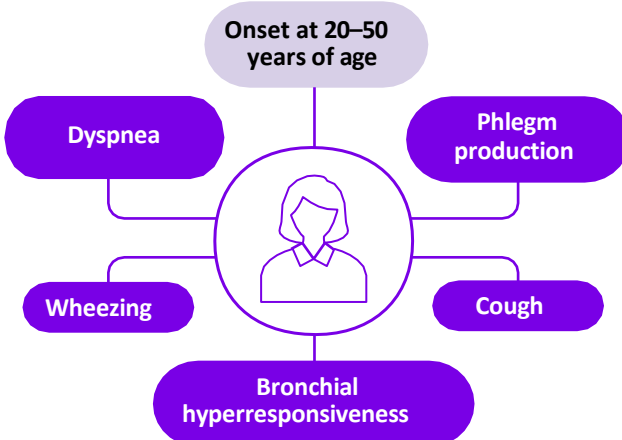


How does AATD lung disease manifest?¹

Accelerated deterioration leading to:

COPD

Emphysema

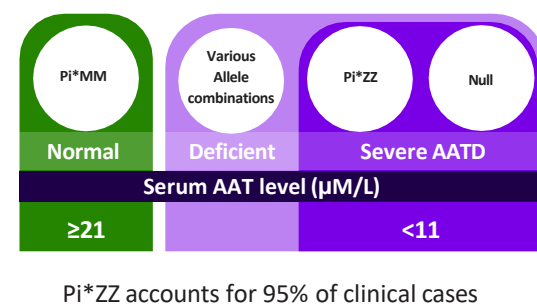


1%–3% of patients with COPD have AATD as the underlying cause^{1,3}

AATD clinical presentation results from both genetic and environmental factors¹

Why does clinical presentation of AATD vary?^{1,2,4-8}

SERPINA1 allelic variation



Environmental factors can accelerate lung damage



Pollution



Cigarette smoking

AATD Underdiagnosis and the Importance of Testing

Delayed diagnosis of AATD can worsen clinical status and impact patient outcomes^{9,10}

Who should be tested for AATD?^{4,11}



Individuals diagnosed with COPD, emphysema, chronic bronchitis, and/or incompletely reversible asthma



Individuals diagnosed with unexplained CLD, necrotizing panniculitis, granulomatosis with polyangiitis, or unexplained bronchiectasis



Biological relatives of individuals diagnosed with AATD

How is AATD diagnosed?¹¹

Confirmatory tests*



SERPINA1 genetic test



Serum AAT level test

*Ensure confirmatory tests are done in parallel.



Up to 95% of individuals with AATD are undiagnosed³

Why is AATD underdiagnosed?^{3,9,12,13}



Testing by serum AAT levels alone is unreliable under certain physiological conditions and patients can be missed.



Nonspecific symptoms (e.g., emphysema, bronchitis, or asthma)



Varied presentation with a range of overlapping symptoms



Lack of awareness, especially in patients without known family history or obvious disease



Median diagnostic delay is 5–8 years from symptom presentation¹⁴

Why is early diagnosis important?^{9,12}

Each additional year of delay in diagnosis results in worsening of



- Predicted % FEV₁ by 0.3%
- SGRQ score
- CAT score

Early diagnosis



Appropriate evaluation and management



Encourage healthy habits, lifestyle and environmental adjustments

AAT, alpha-1 antitrypsin; AATD, alpha-1 antitrypsin deficiency; CAT, COPD Assessment Test; CLD, chronic liver disease; COPD, chronic obstructive pulmonary disease; FEV₁, forced expiratory volume in one second; Pi, protease inhibitor; SGRQ, St George Respiratory Questionnaire.

References

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AATD Diagnostic Testing Options

Notice: Some of the laboratories offering *SERPINA1* genetic testing are listed below. There may be other diagnostic or genetic 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. Sanofi does not review or control the content of non-Sanofi websites. 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 laboratory to confirm test availability, sample shipping information, and all other logistics. This listing does not constitute an endorsement by Sanofi of information provided by any other organizations.

Targeted AATD Testing

Laboratory	Test Name	Test Type	Kits	Sample Type	Billing	Avg TAT	Contact
ARUP Laboratories	AAT (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to AAT Phenotype	AAT protein level, AATD phenotyping	No	Serum: 0.5mL plus WB: 3mL EDTA (lavender or ACD A or B (yellow))	Inst, Ins, self-pay	2-10 days	P: 1-800-522-2787; E: clientservices@aruplab.com W: www.aruplab.com
Blueprint Genetics	SERPINA1 single gene test	Full gene sequencing	Yes	WB: 1mL EDTA (lavender) DNA: min 2µg in TE buffer Saliva swab: per kit	Ins, Inst, self-pay	21-28 days (3-4 weeks)	P: +1 (650) 452-9340; E: support@blueprintgenetics.com W: www.blueprintgenetics.com
Fulgent genetics	AATD (SERPINA1 Single Gene Test) (Sequencing + Del/Dup)	Full gene sequencing	Yes	WB: 4mL EDTA (lavender) or ACD A or B (yellow) DNA: 3µg in EB buffer or equivalent Saliva swab: per kit	Ins, Inst, Self-Pay	3-5 weeks (STAT avail)	P: (626) 350-0537 E: info@fulgentdiagnostics.com W: www.fulgentgenetics.com
Grifols	AlphaID™ Confirm AlphaID™	AAT protein level + genotyping 14 variants Genotyping 14 variants	Yes	Dried blood spot kit Cheek swab kit	*Sponsored	5 days	P: 1-888-474-3657; E: customer.service@grifols.com W: www.alphaID.com
Labcorp	AATD Profile AATD, DNA Analysis	AAT protein level + genotyping 2 variants (S; Z) + PI typing Genotyping 2 variants (S; Z)	Yes (buccal)	Serum: 3mL serum (in 2 1.5mL tubes) plus WB: 7mL EDTA (lavender) or buccal swab WB: 7mL EDTA (lavender)	Inst (acct req'd)	6-12 days	P: 800-345-4363 W: www.labcorp.com
Mayo Clinic Laboratories	SERPINA1 Gene, Full Gene Analysis AAT Proteotype S/Z, LC-MS/MS, serum	Full gene sequencing AAT protein level + proteotype (S; Z only)	Yes	WB: 3 ml EDTA (lavender) or ACD (yellow) Serum: 1.25 mL	Ins, Inst (acct req'd), self-pay	7-14 days 7-14 days	P:800-533-1710; E: mcl@mayo.edu W: www.mayocliniclabs.com
National Jewish Health Advanced Diagnostic Laboratories	AAT Levels, Genotype, and Phenotype	AAT protein level, genotyping 22 variants	No	WB: 2mL ACD A or B (yellow); or EDTA (lavender)	Inst	7-10 days	P: 800.550.6227 E: clinreflabs@njhealth.org W: www.nationaljewish.org/for-professionals
Prevention Genetics, part of Exact Sciences	SERPINA1 via Custom Panels testing	Full gene sequencing	Yes	WB: 3-5mL EDTA (lavender) or ACD A or B (yellow) DNA: 5 – 10 µg at least 100ng/uL	Ins, Inst, Self-Pay	3 weeks	P:(715) 387-0484 ; E: support@preventiongenetics.com W: www.preventiongenetics.com
Quest Diagnostics	AAT Mutation Analysis (15340) AAT Quantitation and Mutation Analysis (17307)	Genotyping 2 variants (S; Z) AAT protein level + genotyping 2 variants (S; Z)	Yes	WB: 2mL EDTA (lavender), heparin (green) or ACD (yellow) Serum: 1mL plus WB: 5mL EDTA ((lavender) or ACD A or B (yellow))	Inst (acct required)	6-10 days	P: 1-866-436-3463 W: www.questdiagnostics.com
Alpha-1 Coded Testing (ACT) Study at University of Florida Health	ACT Research Study Testing	Genotyping and full sequencing	Yes	Dried blood spot kit mailed to subject with consent form	*Sponsored	Call to discuss	P: 1-855-476-1227 E: alpha1lab@alphaone.ufl.edu W: https://redcap.ctsi.ufl.edu/redcap/surveys/?s=3L7JMEW47E

Multi-Gene Panels

Laboratory	Test Name	# Genes	Kits	Sample Type	Billing	Avg TAT	Contact
Blueprint Genetics	Cystic Lung Disease Panel Comprehensive Pulmonary Panel	163 114	Yes	WB: 1mL EDTA (lavender) DNA: min 2µg in TE buffer Saliva swab: per kit	Ins, Inst, Self-Pay	21-28 days (3-4 weeks)	P: (650) 452-9340; E: support@blueprintgenetics.com W: www.blueprintgenetics.com
Fulgent Genetics	Lung Disorders NGS Panel Cystic Lung Disease NGS Panel	69 8	Yes	WB: 4mL EDTA (lavender) or ACD A or B (yellow) DNA: 3µg in EB buffer or equivalent Saliva swab: per kit	Ins, Inst, Self-Pay	3-5 weeks (Rush/STAT available)	P: (626) 350-0537 E: info@fulgentdiagnostics.com W: www.fulgentgenetics.com
Greenwood Genetic Center Laboratories	Comprehensive Pulmonary NGS Panel	147	Yes	WB: 3-4mL EDTA (lavender)	Ins, Inst, Self-Pay	8 weeks	P: 888-GGC-GENE E: labgc@ggc.org W: www.ggc.org
Revvity Omics	Comprehensive Pulmonary Disease Panel	52	Yes	WB: 3-5mL EDTA (lavender) DBS: all spots Saliva: Oragene kit	Ins, Inst, Self-Pay	2-3 weeks	P: 866-354-2910 E: genomics@revvity.com W: www.revvity.com
The Lantern Project (performed at RevvityOmics)	Interstitial Lung Disorders Panel	56	Yes	WB: 3-5mL EDTA (lavender) DBS: all spots Saliva: Oragene kit	*Sponsored	2-3 weeks	P: : 866-354-2910 E: genomics@revvity.com W: www.LanternProjectDx.com

*Sponsored testing is performed at no charge; local charges may apply for sample collection, processing or shipping

Avg TAT, average turnaround time; DBS, dried blood spot; Ins, Insurance; Inst, Institutional billing; WB, whole blood.