

It wasn't just his emphysema. He was also

BORN WITH IT

CHRIS, 66
LIVING WITH AATD
DIAGNOSED AT 58

Photo is representative and not an actual image of Chris as a baby.

Alpha-1 antitrypsin deficiency (AATD) is the best-known genetic risk factor for COPD¹

Are you managing 100 patients with COPD in your practice? If so, **you statistically have a patient with AATD**, a condition caused by pathogenic variants in the *SERPINA1* gene.²⁻⁴

In the United States, severe AAT deficiency^a is estimated to affect **nearly 100,000 individuals**.^{5,6} However:



>90%
of individuals
with AATD are
undiagnosed⁷



Due to the
overlapping clinical
presentations, AATD
is often **incompletely
diagnosed** as COPD.⁸

^a≤11 μM

Diagnostic delay leads to lung function decline, worsening QoL, and decreased survival^{9,10}

Each additional year of
delay is associated with^{9,a}:

**1.6
POINTS**

worse SGRQ score^{b,c}

**0.7
POINTS**

worse CAT score^d

Earlier detection and diagnosis can improve survival^{10,e}

A longer diagnostic delay is associated with worse overall survival, as observed in a multivariable analysis (n=229).

**61%
HIGHER RATE OF MORTALITY**

during follow-up associated with
each proportional (~2.7-fold) increase
in diagnostic delay.

(HR 1.61; 95% CI 1.09–2.38; *P*=0.016)

^aData from 40 newly diagnosed patients with AATD. Included 31 patients (78%) with ZZ genotype and 9 patients (22%) with SZ genotype. At the onset of the study, 4 patients (10%) were current smokers, and 21 patients (58%) were ex-smokers. ^b*P*<0.001; ^cThe SGRQ is a commonly used measure of QoL, specifically designed for patients with obstructive airway diseases. ^d*P*=0.004; ^eLonger diagnostic delay was independently associated with worse overall survival independent from age, smoking status, BMI, FEV₁, and long-term oxygen treatment.

AATD creates a broad, multidimensional burden for patients¹¹⁻¹⁶

AATD is often overlooked in patients with classic symptoms of other chronic pulmonary conditions, such as irreversible asthma, emphysema, and unexplained bronchiectasis.^{2,13}

Symptoms include¹³:



Shortness of breath during daily activities



Wheezing



Decreased ability to exercise



Excessive mucus production while coughing

Risk of accelerated lung decline is exacerbated by factors such as a patient's environment (eg, pollutants, respiratory infections) and lifestyle (eg, smoking).

Genetic and serum level testing are the only ways to rule out or diagnose AATD^{8,17}

Parallel testing is recommended for the diagnosis of AATD^{18,19}:

- **Genetic testing** confirms a diagnosis of AATD and determines the specific genotype^{18,20}
- **Testing AAT levels** determines whether protein deficiency is present and assesses severity^{17,20}

ATS, CTS, ERS, GOLD, and others recommend testing in the following populations^{13,14,21,22}:

- **All patients with a diagnosis of COPD**
- **Symptomatic individuals:**
 - Adult-onset asthma (persistent, incompletely reversible airflow obstruction)
 - Unexplained bronchiectasis
 - Unexplained chronic liver disease
 - Necrotizing panniculitis
 - Granulomatosis with polyangiitis (GPA)
- **Asymptomatic individuals** with persistent airflow obstruction, especially those with identifiable risk factors (eg, heavy smoking history, exposure to air pollution, or occupational inhalants)
- **First-degree relatives** of patients diagnosed with AATD

Learn how to detect AATD in your patients at DiagnoseAATD.com



AAT=alpha-1 antitrypsin; **AATD**=alpha-1 antitrypsin deficiency; **ATS**=American Thoracic Society; **BMI**=body mass index; **CAT**=COPD Assessment Test; **COPD**=chronic obstructive pulmonary disease; **CTS**=Canadian Thoracic Society; **ERS**=European Respiratory Society; **FEV₁**=forced expiratory volume in 1 second; **GOLD**=Global Initiative for Chronic Obstructive Lung Disease; **QoL**=quality of life; **SGRQ**=St. George's Respiratory Questionnaire.

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