

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 COPD[1]

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 undiagnosed[7]



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)

[a]Data 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; [c]The SGRQ is a commonly used measure of QoL, specifically designed for patients with obstructive airway diseases. [d]*P*=0.004; [e]Longer diagnostic delay was independently associated with worse overall survival independent from age, smoking status, BMI, FEV_1, and long-term oxygen treatment.

AATD creates a broad, multidimensional burden for patients[11-16]

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 include[13]:


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; **FEV_1=**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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