Detecting Alpha-1 Antitrypsin Deficiency (AATD)

Signs & Symptoms

may have

ALPHA-1 ANTITRYPSIN DEFICIENCY (AATD)

a life-limiting, rare genetic disease

PATIENTS PRESENTING WITH

- **frequent infections**
- **chronic cough**
- **wheezing**
- **shortness of breath with exertion**
- **increased phlegm production**
- **swelling of the abdomen (ascites)**
- **vomiting blood or blood in the stool; dark urine**
- **yellowing of the eyes and skin**

Alpha-1 antitrypsin protein (AAT) is **produced by the liver and protects the lungs from inflammation caused by infection or inhaled irritants.** AATD occurs when there is a **deficiency of AAT in the blood,** increasing the risk of lung and liver disease.

As **symptoms** are similar to other lung and liver diseases, **early diagnosis is a challenge.** Numerous patients remain undiagnosed. Without appropriate intervention and care, **patients may have a decreased life expectancy,** progressing from the need for:

Home Care → **Oxygen Therapy** → **Mobility Support** → **Transplantation.**

Re-occurring hospital admissions and absenteeism from school or work may also be **signs that your patient should be tested for AATD.**

Early diagnosis and timely treatment are essential to slow the progression of organ deterioration and preserve lung tissue.

Who to test for AATD?

Consider testing for AATD for your patients with:

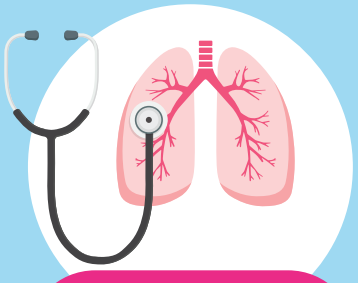
- COPD
- a family history of AATD
- bronchial asthma
- bronchiectasis
- emphysema
- a diagnosis of adult onset asthma
- little to no history of smoking
- the need for a lung and/or liver transplant (patients on waiting lists)
- chronic liver disease
- hepatitis and liver cirrhosis
- unexplained liver disease
- hepatocellular carcinoma
- panniculitis
- vasculitis

Testing all COPD/ asthma patients for AATD is a simple step you can take to ensure early diagnosis.

How to test for AATD?

Diagnostic steps[1] for healthcare professionals

AATD is easily **diagnosed through a simple blood draw** or **finger prick sample.** If the AAT level is lower than normal, **a follow-up genotype or a phenotype blood test** can be ordered, in addition to the following tests:



Full physical examination

CT of the lungs or chest X-Ray

Pulmonary function test

Liver function test

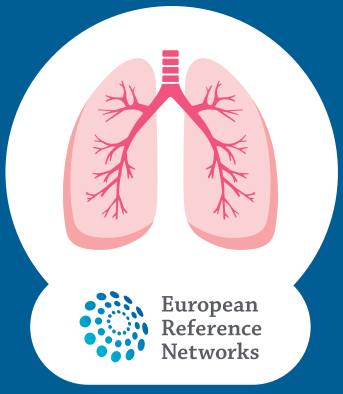
Next Steps[1]

upon identification that your patient may have AATD:

Referral to a centre of excellence

All tests should be **forwarded to a centre of excellence for further evaluation and confirmation** of diagnosis by a specialist with expertise in AATD.

Connecting with your national centres of excellence supports coordinated care delivery for these complex patients, while enhancing knowledge exchange within your medical community.



ERN Lung[2]

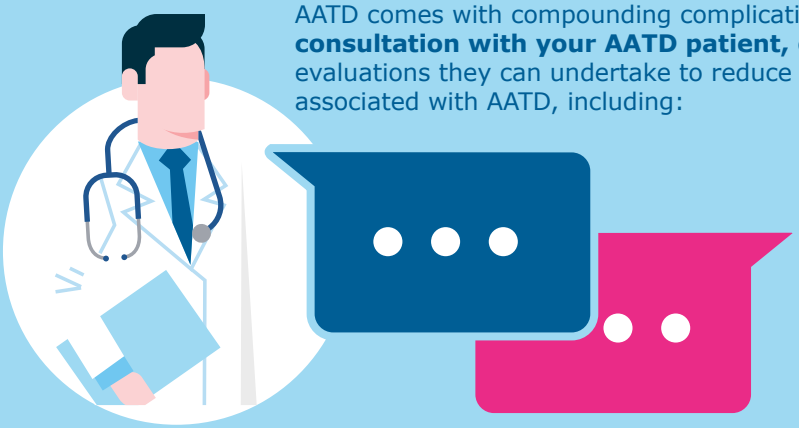
- AATD specific branch: the AATD Core Network
- Cross-border blood sample exchange for the detection of rare mutants of the AAT gene
- Data repository for all respiratory disorders, including disease-specific registries

ERN Rare-Liver[3]

- Inclusion of AATD in the Network on Metabolic, Biliary Atresia & Related Disease and linkage with the European Alpha-1 Liver Study group
- Collecting key patient outcome data to monitor quality standards
- Developing best practice patient information leaflets

[1] 2017, Alpha-1 European Expert Group Recommendations. Alpha-1 Global: www.alpha-1global.org
[2] www.ern-lung.eu
[3] www.rare-liver.eu

Continuous monitoring & follow-up[1] of AATD



AATD comes with compounding complications and comorbid conditions. **During your next consultation with your AATD patient,** consider discussing the steps they can take and evaluations they can undertake to reduce the progression of lung, liver or skin diseases associated with AATD, including:

- **evaluation by a pulmonologist**
- **evaluation by a liver specialist**
- **treatment options and benefits**
- **vaccinations for influenza, pneumococcal, hepatitis A/B**
- **lifestyle factors, such as stress, alcohol consumption, smoking (where lifestyle modifications may help), occupational and environmental risks**

Active management and treatment of complications will be ongoing with your AATD patient.

Due to the **hereditary nature** of AATD, continue to monitor symptoms presented by their family members and refer accordingly.

Key facts & Figures Section

AATD is the most **common hereditary condition** in adults worldwide[4]. The prevalence of its severe form varies across Europe, affecting about

1/1,500

to 3,500 individuals, whereas mild forms are much more prevalent[5].

Although approximately

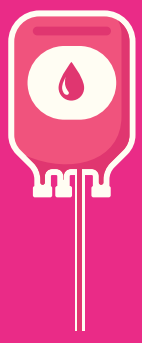
120.000[6]

people in Europe carry the PI*ZZ genotype associated with AATD, **only a small proportion have been diagnosed and receive treatment.**

Alpha-1 is the **most widely recognised rare, genetic cause of chronic obstructive pulmonary disease** (COPD)[7]. More than **66 million people have COPD** in the European region, of which approximately **2 million cases are caused by AAT deficiency.**[7]

50 YEARS

Alpha-1 antitrypsin deficiency was **discovered more than 50 years ago,** but much remains unknown.[7]



While there is currently no cure, **treatments are available** to better manage the disease, treat symptoms, and slow down the progression of organ damage, such as **plasma-derived therapy** for Alpha 1 patients.

[4] Gramegna, Andrea et al. "Alpha-1 antitrypsin deficiency as a common treatable mechanism in chronic respiratory disorders and for conditions different from pulmonary emphysema? A commentary on the new European Respiratory Society statement" Multidisciplinary respiratory medicine vol. 13 39. 8 Oct. 2018, doi:10.1186/s40248-018-0153-4

[5] 2017, Alpha-1 European Expert Group Recommendations. Alpha-1 Global: www.alpha-1global.org

[6] Campbell, Edward (the reference number one of the Alpha 1 European Expert group recommendations)

[7] 2017, Alpha-1 European Expert Group Recommendations. Alpha-1 Global: www.alpha-1global.org

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