Detecting Alpha-1 Antitrypsin Deficiency (AATD)

**Signs & Symptoms**

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.

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

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:

- 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

**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
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. 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.

Although approximately 120,000 people in Europe carry the PI*ZZ genotype associated with AATD, only a small proportion have been diagnosed and receive treatment.

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

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.

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

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