PROVIDER INFORMATION SHEET: ALPHA 1 ANTITRYPSIN DEFICIENCY

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Overview

Alpha 1 antitrypsin deficiency (A1AD) is due to a reduced level of alpha-1 antitrypsin (AAT), a protein that protects the lungs and liver. **Which genetic change a person has**, along with certain environmental factors, impacts the potential to develop health concerns.

- The **M variant** produces **normal** levels of AAT.
- The **S variant** produces **reduced** levels of AAT.
- The **Z variant** produces **very little** AAT.

Potential Health Concerns

- **Having ZZ (two Z variants)**
  - Primary features in **adults** include cirrhosis, liver fibrosis, and COPD.
  - **2 to 10% of children** develop neonatal cholestasis**.** Emphysema is extremely rare.
  - Disease expression varies within and between family members.
- **Having SZ (one S and one Z variant)**
  - Increases the risk of emphysema and COPD in adults. The risk is higher for smokers or those exposed to dust and pollutants.
- **Having SS (two S variants) or MS (one M and one S)**
  - **Is not believed** to pose a significant health risk.

When to suspect?

- In people with COPD, unexplained chronic liver disease, and/or panniculitis.
- In those with relatives who have been diagnosed with A1AD.

Possible Next Steps

- Diagnosis: low serum concentration of AAT **AND** either a pathogenic variant in each *SERPINA1* gene **OR** detection of a functionally deficient AAT protein variant.
- Refer to a
  - Genetic counselor to review, order, and discuss the results of genetic testing options.
  - Pulmonologist to assess lung function and imaging and to manage COPD/emphysema.
  - Hepatologist/Gastroenterologist to evaluate liver function and manage hepatic issues.

Resources

- Alpha-1 Foundation
- GeneReviews – AATD