

Alpha 1 antitrypsin

General:

Alpha-1-antitrypsin is an *acute phase protein*; its function is the inhibition of the activity of proteolytic enzymes. Thus, it is a protease inhibitor. It is synthesized in alveolar macrophages, in monocytes and in hepatocytes. The highest inhibitory activity is against elastase released by granulocytes. In case of a defect the non-inhibited elastase activity leads to increased reduction of the structural alveolar elastin and subsequently to emphysema. Acquired AAT defects are caused by oxidation from cigarette smoke thereby inactivating AAT.

Clinical significance: Early diagnosis of reduced alpha-1-antitrypsin as well as phenotyping. Alpha-1-antitrypsin in stool is used as a marker for protein loss in chronic inflammatory diseases of the intestine (e.g. Crohn's disease). An increase of the fecal proteinase inhibitor concentrations is caused by an increased permeability of intestinal epithelium. The reason is an abnormal arrangement of the intestinal "tight junctions". This leads to an enteral loss of plasma alpha-1-antitrypsin. Protein-losing enteropathies are found in Crohn's disease, colitis ulcerosa, parasites, bacteria, viruses or fungi, bacterial overgrowth of the small intestine (overgrowth syndrome), food allergies and other allergies, celiac disease, lymph drain obstruction (intestinal lymphomas, mesenteric ischemia etc.). Increased alpha-1-antitrypsin values are observed in long-term treatment with non-steroidal antiinflammatories (NSAID). Other markers of permeability malfunction are: PMN-elastase (migration of leukocytes), IgA (immune reaction of the intestinal wall by lymphocytes) and albumin (plasma passage into the intestinal lumen).

The following tests are available:

- **Alpha-1-antitrypsin in serum**

Indication: suspicion of hereditary A1-AT-deficiency in combination with the following symptoms: icterus prolongatus in the newborn, hepatitis of unclear genesis in infancy and childhood, pulmonary emphysema of the adult, hepatitis or hepatocirrhosis of unknown origin in the adult.

Material: 1 ml serum

TAT: 7-10 days*

Method: nephelometry

Units: mg/dl

Ref.- range: 90 - 200

decrease of the alpha-1-fraction in the serum electrophoresis can point to AAT

Note: deficiency

- **Alpha-1-antitrypsin in stool**

Indication: suspicion of chronic inflammatory intestinal disease

Material: 5 g stool

TAT: 7-10 days*

Method: EIA

Units: $\mu\text{g/g}$ stool

Ref.- range: <268

- **Alpha-1-antitrypsin genotyping**

Indication: Lung emphysema, liver disorders, icterus prolongatus, differentiation of degraded AAT, genetic carrier testing

Material: 5 ml EDTA-blood

TAT: 10-14 days*

Method: PCR

- **Alpha-1-antitrypsin phenotyping**

Indication: Differentiation of decreased AAT, suspicion of genetic carrier, prolastin therapy monitoring.

Material: 3 ml serum

TAT: 10-14 days*

Method: IEF

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit <http://www.fml-dubai.com/parameter-listings/>