:: Alpha-1 antitrypsin deficiency

- *This document is a translation of the French recommendations drafted by Dr Mornex and Lachaux, reviewed and published by Orphanet in 2010.*
- *Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.*

Synonyms
AAT deficit

Definition:
Autosomal recessive genetic disorder, characterised by a reduction in the serum level of alpha-1 antitrypsin (AAT), an inhibitor of elastase activity. In adults, this disorder manifests itself primarily as **pulmonary emphysema** (in which case patients receive conventional bronchodilator treatment and some receive replacement therapy); very rarely, it manifests itself as panniculitis, and in children, **liver disease**.

Further information:
See the Orphanet abstract

Pre-hospital emergency care recommendations
Call for a patient suffering from alpha-1 antitrypsin deficiency

Synonyms

- AAT deficit

Definition

- autosomal recessive genetic disorder characterised by a reduction in the serum level of alpha-1 antitrypsin (AAT, an inhibitor of elastase activity); in adults, it manifests itself primarily as pulmonary emphysema; in children, it is manifested in the form of liver disease

Specific risks in emergency situations

- pneumothorax
- exacerbation of chronic bronchitis

Commonly used long-term treatments

- bronchodilators
- rarely, replacement therapy with human AAT

Complications

- no specific complications

Specific medical care prior to hospitalisation

- there is no specific therapeutic information on the management of complications: follow the usual recommendations

For further information

- Please visit www.orpha.net and type the name of the disease -> in the summary page click on "Expert centres" on the right tab -> select "United Kingdom" in the "Country" field in the Expert centres page.

Recommendations for hospital emergency departments

Emergency situations

- Exacerbation of chronic bronchitis accompanying emphysema
- Pneumothorax

Immediate diagnostic and therapeutic measures

- In terms of clinical manifestations and management of complications, there is no specific information relating to AAT deficiency. **Follow the usual recommendations**

Drug interactions

- No specific drug interactions in respect of medicinal products used in emergency medicine or long-term treatment

Precautions for anaesthesia

- No specific precautions

Additional therapeutic measures and hospitalisation

- **Admission**: infants with jaundice should preferably be admitted to a single room since they are vulnerable
- **Accompanying family members**: a mother-and-child room is desirable and psychological support should be offered
- **Inform the patient** and/or the parents about the various possible outcomes of the disorder, the precautions to be taken and things that must be avoided. Further information can be obtained from Alpha1 Awareness UK, (http://www.alpha1awareness.org.uk/welcome.htm).

Organ donation

- Yes, organ donation is possible, apart from liver and lung donation in the case of emphysematous patients

Emergency telephone numbers

- Please visit www.orpha.net and type the name of the disease -> in the summary page click on "Expert centres" on the right tab -> select "United Kingdom" in the "Country" field in the Expert centres page.

Documentary resources

- Silverman EK, Sandhaus RA: **Clinical practice. Alpha 1-antitrypsin deficiency**. *New Engl J Med* 2009, 360: 2749-57.

These recommendations have been compiled in collaboration with Dr. Jean-François Mornex –National reference centre for rare pulmonary disorders, Louis Pradel Cardiovascular and Respiratory Disorders Hospital, Claude Bernard Hospital, Lyon; Dr. Alain Lachaux – Department of Paediatric Gastroenterology, Hepatology and Nutrition, Lyon University Hospitals, Bron; the Alpha-1 Antitrypsin Deficiciency Patients' Association in France and with Dr. Gilles BAGOU – SAMU-69, Lyon.

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