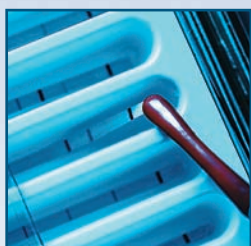


GenoType[®] AAT

Based on DNA•STRIP[®] Technology



- simple
- safe
- fast
- easy to combine
- can be automated



CE-labelling
Quality management
certified to ISO 9001



Alpha-1-Antitrypsin

The protease inhibitor alpha-1-antitrypsin (AAT), found in high concentrations in the plasma, inhibits trypsin and also neutrophil elastase. The presence of AAT deficient alleles leads to a pathological accumulation of AAT in the hepatocytes due to a disturbed protein export from the liver, and hence to a lack of functional inhibitor in the body tissue. The resultant unchecked action of the elastase results in particular in liver damage. Pulmonary emphysema, chronic obstructive pulmonary disease etc. are among the most common symptoms caused by an AAT deficiency. Furthermore, due to the toxic effect of the accumulated AAT on the liver cells, clinical pictures such as cirrhosis of the liver and even liver carcinoma are among other late sequelae. The causes of AAT deficiency are mainly two allele variants of the AAT gene. In contrast to normal alleles (PiM), the risk alleles PiZ and PiS are associated with considerably lower plasma concentrations of AAT. PiZ is by far the most common and diagnostically significant deficient allele (in 95% of all patients with severe AAT deficiency) whereas heterozygous (PiMZ or PiSZ) and homozygous PiS carriers as a rule only fall ill if there are additional risk factors involved such as smoking. With early diagnosis the patient can accordingly avoid negative factors, clinical late sequelae can be prevented or minimised.

With an incidence of 1:2000, AAT deficiency is one of the most common potentially lethal hereditary diseases in Europe. A Scandinavian study identified as much as 5% of the normal population as heterozygous PiZ carriers with a significantly reduced AAT level, thus clearly highlighting the relevance of early diagnosis.

Indications for the GenoType[®] AAT test kit

- Adult patients with abnormal liver values, hepatitis or liver cirrhosis of unclear origin
- Differential diagnosis with COPD, pulmonary emphysema or asthma
- Patients with clinically diagnosed AAT deficiency
- Members of index families
- For the differential diagnosis of hepatitis and hepatic dysfunction of unclear origin in infants and young children and also prolonged neonatal jaundice.

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The **GenoType® AAT** test kit identifies the clinically relevant PiZ and PiS mutations and also the normal PiM allele safely, quickly and reliably in a single procedure. Thanks to the simple combining of **GenoType® AAT** with all the parameters of the **GenoType®** series, it can easily be integrated into the routine diagnostics of any laboratory.

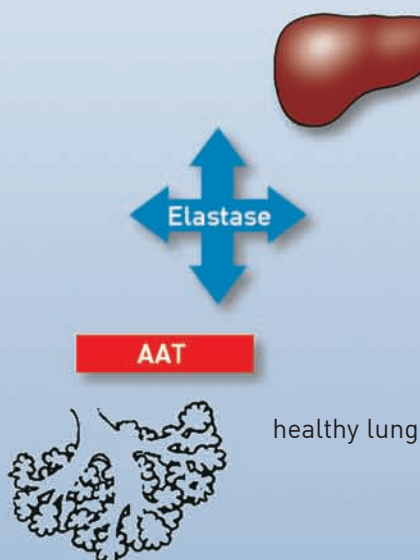
For technical information, please see the brochure "**DNA•STRIP® Technology**".
Further literature is available direct from HAIN LIFESCIENCE.

Key words

Genotype	Allele Frequency	Trypsin Inhibition	Risk Classification	Risk Classification (Smokers)
MM	90 %	100 %	no risk	slight risk
MZ	4 %	61 %	slight risk	increased risk
MS	n.k.	83 %	slight risk	increased risk
SS	0,1 %	63 %	no risk	slight risk
SZ	0,12 %	38 %	moderate risk	high risk
ZZ	0,04 %	15 %	high risk	very high risk

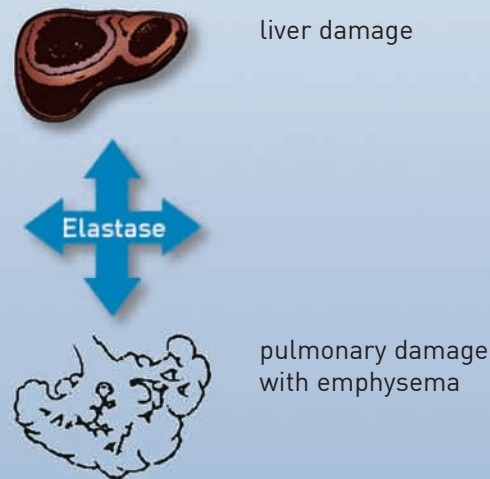
Wildtype:

- Normal AAT export from liver
- Protection against elastase especially in the lung



AAT-Mutation:

- Disturbed AAT export from liver
- Reduced concentration of functional AAT in the lung
→ insufficient protection against elastase
- Pathological accumulation in the liver



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