

Alpha-1 antitrypsin deficiency ...



... A risk for lung and liver!

Your molecular genetic test system for reliable detection of the two major deficiency alleles of the alpha-1 antitrypsin gene.



Your benefits of using GenoType AAT

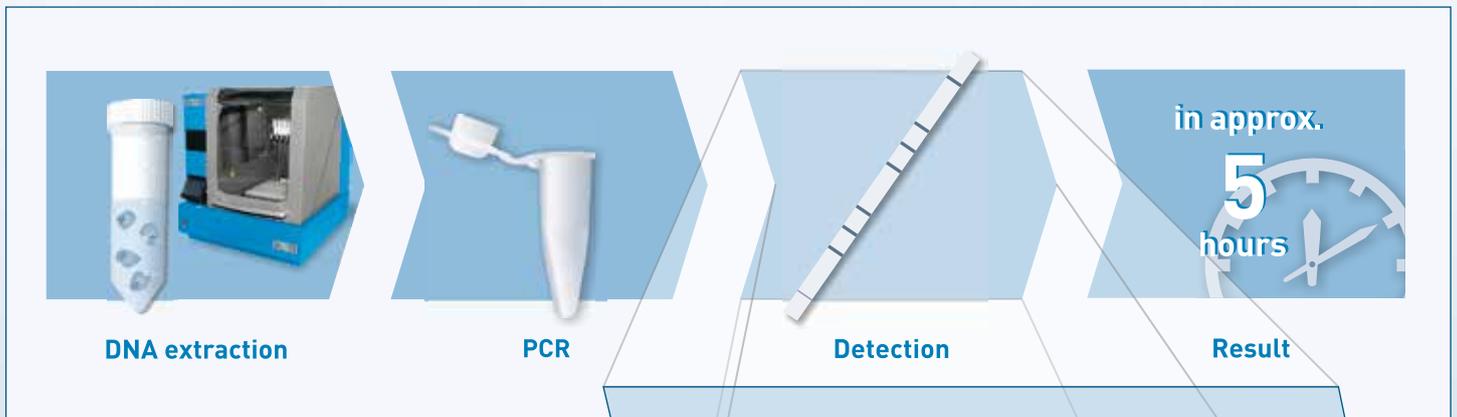
- **Certainty:** Genotyping with **GenoType AAT** generates reliable evidence about the existence of a genetic AAT deficiency.
- **Reliable results:** An internal control guarantees reliable test results at any time.
- **User-friendly:** The user-friendly **DNA•STRIP** technology combines both high information content and efficient processing. A ready-to-use amplification mix including the Taq polymerase is provided in the kit, this saves time and money.
- **Comprehensive diagnostics:** We offer a broad range of test systems based upon the same technology. Thus, the simultaneous processing of different human genetic parameters allows an optimal integration into your routine laboratory diagnostics.
- **CE-IVD certified:** No need for elaborate validation studies.

Facts

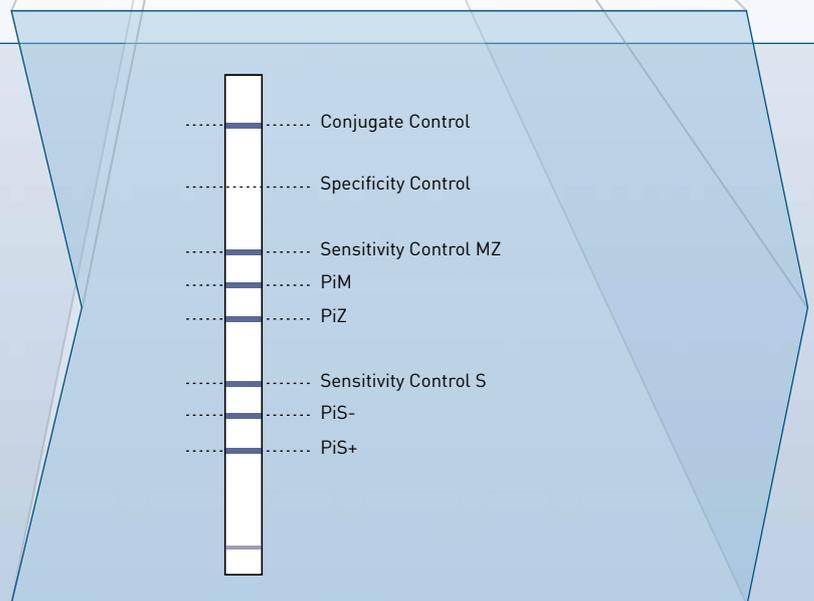
Genetic alpha-1 antitrypsin deficiency is one of the most frequent inherited disorders in Europe. Alpha-1 antitrypsin (AAT) is produced in the liver and transported via the bloodstream. It plays a central role in the protection of the lung against excessive digestion of proteins by proteases. Thus, AAT controls the activity of neutrophil elastase, which non-specifically destroys proteins in the context of the immune response. As a result of an AAT deficiency, the enzyme activity is not controlled and therefore, lung tissue gets destroyed. This can lead to severe damages, for example in the form of chronic obstructive pulmonary disease (COPD). A reduced release of AAT into the bloodstream results in AAT deficiency in the lung as well as in AAT accumulation in the liver. This accumulation leads to liver diseases which increase the risk for liver cirrhosis and liver tumors.

Certain gene defects are the leading cause of AAT deficiency in the lung. A distinction is made between the normal allele PiM and the two risk alleles PiZ and PiS. The most frequent type of genetic defect is the homozygote Z phenotype (PiZZ), which is associated with significantly reduced serum concentration of AAT. The homozygote S phenotype (PiSS) leads, as well as a combination of both risk alleles (PiSZ), to a decreased plasma concentration of AAT in the lung. Genetic testing is recommended particularly in case of young patients with a chronic cough or shortness of breath.

GenoType AAT: Reliable detection of the two major deficiency alleles of the AAT gene



GenoType AAT is based on the user-friendly **DNA•STRIP** technology: The isolated DNA is amplified and detected via reverse hybridization and an alkaline phosphatase reaction on a membrane strip. Thus, mutations of the AAT gene, which lead to the Z and S phenotypes, are detected fast and reliably. An evaluation template and an interpretation table facilitate the result interpretation.



Further information is directly available from Hain Lifescience or your local distributor.

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