gb GENETIC A1AT

Clinical implications

Alpha-1-antitrypsin (A1AT) is one of the serine protease inhibitors (SERPIN) which serves as an inhibitor of neutrofile elastase in lungs. The mutations in the A1AT gene lead to changes in the protein structure and loss of function. Alpha-1-antitrypsin deficiency is associated with lung diseases such as chronic obstructive pulmonary disease (COPD). The main risk factors for COPD are smoking, dusty work or repeated respiratory infections. A1AT deficiency can cause liver disease (hepatitis, cirrhosis) in both, neonates and adults. The most frequent mutations which cause a deficiency in alpha-1-antitrypsin are PI*S and PI*Z. Mutation PI*S is caused by substitution of E264V and makes a gradual reduction in protein levels. Mutation PI*Z is caused by substitution of E342K and makes a significant A1AT level reduction in blood serum.

Principle of detection

The kit is intended for detection of mutations PI*S and PI*Z in the alpha 1 antitrypsin gene in human genomic DNA. Detection is based on **real-time polymerase chain reaction (qPCR) using fluorescently labelled probes (allelic discrimination)**.

Available products

Cat. No.	Product	rxn
3251-025	gb GENETIC A1AT	25
3251-050	gb GENETIC A1AT	50

1 kit contains reagents to provide 25 or 50 PCR reactions (20 µl volume of each reaction).

Parameters of the diagnostic kit

- ***in vitro* diagnostics**
- **CE IVD, IVDR certified**
- **ready-to-use assay**
- **sample concentration 10-100 ng/µl**
- **positive and negative controls included**
- **FAM and HEX channels detection**
- **identical amplification profile as gb HEMO, gb GENETIC, gb PHARM kits**

Content of the diagnostic kit

*	Component	Conc.	Purpose
	Assay qPCR A1AT (PI*S)	1.25×	Detection assay
	Standard WT A1AT (PI*S)	10^4 cop/µl	Positive Control
	Standard MUT A1AT (PI*S)	10^4 cop/µl	Positive Control
	Standard HET A1AT (PI*S)	10^4 cop/µl	Positive Control
	Assay qPCR A1AT (PI*Z)	1.25×	Detection assay
	Standard WT A1AT (PI*Z)	10^4 cop/µl	Positive Control
	Standard MUT A1AT (PI*Z)	10^4 cop/µl	Positive Control
	Standard HET A1AT (PI*Z)	10^4 cop/µl	Positive Control
	Deionized Water		Negative Control

** Lid colour*



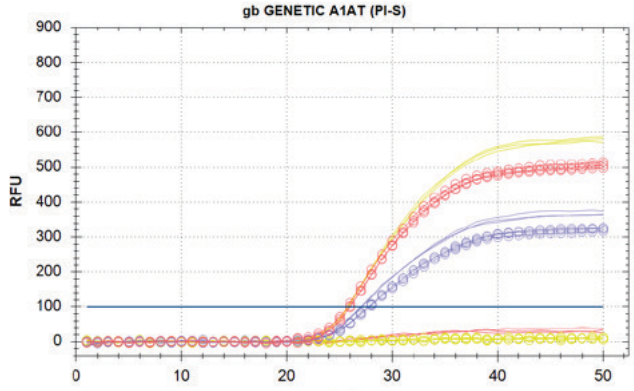


*Fig. 1 – Detection of A1AT (PI*S) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel*

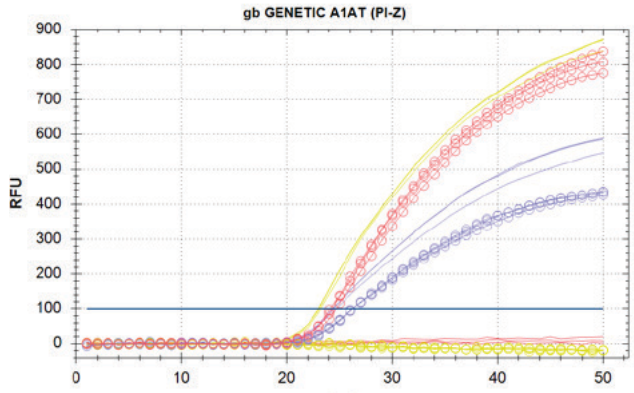


*Fig. 2 – Detection of A1AT (PI*Z) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel*

Validated for cyclers

- Rotor-Gene 3000/6000/Q (Corbett Research Qiagen)
- CFX96/CFX96 Touch (Bio-Rad)
- ABI 7500/7500 Fast (Applied Biosystems)
- MIC (Bio Molecular Systems)
- Light Cycler 480/Cobas z480 (Roche Diagnostics)
- QuantStudio 5 (Applied Biosystems)

CE IVD