Patients with deficient alleles may be more common than you think¹



Early screening with AlphalD[™] can detect deficient alleles

AlphaID screens for the most-prevalent alleles associated with AAT deficiency, also known as alpha-1, including **S**, **Z**, **F**, **I**, rare alleles, and null alleles.²

- 15% of patients screened were positive for at least 1 deficient allele³
- Deficient alleles can be passed on to family members, including children^{4,5}
- The "F" and "I" allele combinations, as well as rare variants, were found in greater than 1% of more than 1 million patients screened²

For more information, please contact your Grifols sales representative



Scan here to order AlphalD[™]

The receipt of this free testing service does not create any expectation or obligation to purchase or use any product or service offered by any manufacturer.



GRIFOLS

AlphalD[™] screens for 98% of all alleles found in people with AAT deficiency²

Below is an overview of the alleles that are identified with AlphaID[™]

Allele type	Genetic risk of lung disease?	Genetic risk of liver disease?	Details
М	No	No	The family of normal alleles is referred to as M ^{6,7}
S	Yes	No	S produced moderately low levels of alpha1-antitrypsin (AAT) ^{6,8-10}
Z	Yes	Yes	Z produced very little AAT ⁶
F	Yes	No	F is dysfunctional as it produces a normal quantity of AAT protein, but the protein does not function properly ⁶
I	Yes	No	I is dysfunctional as it produces a normal quantity of AAT protein, but the protein does not function properly ^{6,8}
M Malton	Yes	Yes	M-like deficient allele with serum AAT levels approximately 10% to 15% of normal AAT levels ^{8,11}
M Palermo	Unknown	Unknown	M-like deficient allele. Insufficient data available to determine genetic risk ⁸
M Nichinan	Yes	Unknown	M-like deficient allele which undergoes a conformational change with a consequent decrease in serum AAT levels ^{8,12}
M Procida	Yes	Unknown	M-like deficient allele which has mildly reduced inhibitory activity, similar to that of the S allele ^{8,10,13}
M Heerlen	Yes	No	M-like deficient allele with an increased genetic risk of lung disease ^{8,10}
P Lowell	Yes	Unknown	PI*P allele with an increased genetic risk of lung disease ^{8,10}
P Duarte	Yes	Yes	PI*P allele with an increased genetic risk of lung disease8
Y Barcelona	Yes	Unknown	Less frequent allele with an increased genetic risk of lung disease ^{14,15}
S liyama	Yes	Yes	Less frequent allele with an increased genetic risk of both lung and liver disease $^{\!\!^{6,8}}$
Q0 Granite Falls	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 West	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 Bellingham Falls	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 Mattawa	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 Ourem	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 Clayton	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰
Q0 Saarbruecken	Yes	No	NULL allele can result in either no mRNA product or no protein production ¹⁰

References: 1. de Serres FJ. *Environ Health Perspect*. 2003;111(16):1851-1854. 2. Data on file, Alpha-1 Genetics Laboratory, Grifols. 3. Data on file, Total Grifols Testing Program, Grifols. 4. American Thoracic Society/European Respiratory Society. *Am J Respir Crit Care Med*. 2003;168(7):818-900. 5. de Serres FJ et al. *Clin Genet*. 2003;64(5):382-397. 6. Stoller JK et al. In: Adam MP, Everman DB, Mirzaa GM, et al, editors. *GeneReviews*[®] [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. https://www.ncbi.nlm.nih.gov/books/NBK1519/7. Lara B et al. *Respir Res*. 2014;125(15): 1-13. 8. Seixas S et al. *Appl Clin Genet*. 2021;14:173-194. 9. McKinney EF et al. *Semin Immunopathol*. 2014;36:461-478. 10. Foil KE. *Ther Adv Chronic Dis*. 2021;12:33-48. 11. Reid CL et al. *Gastroenterology*. 1987;93:181-187. 12. Matsunaga E et al. *Am J Hum Genet*. 1990;46(3):602-612. 13. Takahashi H et al. *J Biol Chem*. 1988;263(30):15528-15534. 14. Miravittles M. *Respir Med*. 2000;94(suppl 3):S12-S15. 15. de la Roza C et al. *Arch Bronconeumol*. 2006;42(6):290-298.



