AlphaID™ At Home Genetic Health Risk Service: Table of Variants

Detailed description of the 14 variants detected by the AlphaID™ At Home Genetic Health Risk Service in the *SERPINA1* gene.

Variant tested	Associated genetic marker (rs)[1]	Clinical Significance of Variants - ClinVar accession number[2]	Global Minor Allele Frequency (MAF) %[3]	Biological explanation	References[4]
PI*S	rs17580	VCV000017969	2%	The variant is a change from A to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is transported out of the liver less effectively. It causes a **mild plasma deficiency** and a **mild accumulation** of protein in the liver.	1, 3, 12, 13
PI*Z	rs28929474	VCV000017967	0.4%	The variant tested is a change from a G to A in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is not transported out of the liver effectively. It causes a **severe plasma deficiency** and a **severe accumulation** of protein in the liver.	1, 2, 14, 15, 16

Variant tested	Associated genetic marker (rs)[1]	Clinical Significance of Variants - ClinVar accession number[2]	Global Minor Allele Frequency (MAF) %[3]	Biological explanation	References[4]
PI*F	rs28929470	VCV000017961	0.1%	The variant is a change from C to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that has a dysfunctional and reduced activity. It has normal to mild plasma levels. It causes a **mild plasma deficiency without accumulation** of protein in the liver.	17, 18, 19
PI*I	rs28931570	VCV000017974	0.06%	The variant is a change from C to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is transported out of the liver less effectively. It causes a **mild plasma deficiency** and a **mild accumulation** of protein in the liver.	20, 21, 22, 23
PI*M procida	rs28931569	VCV000017971	< 0.001%	The variant is a change from T to C in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is broken down more quickly. It causes a **severe plasma deficiency without accumulation** of protein in the liver.	24, 25, 26, 27
PI*M malton	rs775982338	VCV000315028	< 0.001%	The variant is a deletion of TTC in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is not transported out of the liver effectively. It causes a **severe plasma deficiency** and a **severe accumulation** of protein in the liver.	21, 26, 28
PI*S iiyama	rs55819880	VCV000017992	< 0.001%	The variant is a change from C to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is not transported out of the liver effectively. It causes a **severe plasma deficiency** and a **severe accumulation** of protein in the liver.	29, 30, 31

Variant tested	Associated genetic marker (rs)[1]	Clinical Significance of Variants - ClinVar accession number[2]	Global Minor Allele Frequency (MAF) %[3]	Biological explanation	References[4]
PI*Q0 granite falls	rs267606950	VCV000017976	< 0.001%	The variant is a deletion of a C in the DNA sequence of the *SERPINA1* gene. It results in no production of protein. It causes a **very severe plasma deficiency without accumulation** of protein in the liver.	32, 33
PI*Q0 west	rs751235320	VCV000189064	< 0.001%	The variant is a change from G to T in the DNA sequence of the *SERPINA1* gene. It results in no production of protein. It causes a **very severe plasma deficiency without accumulation** of protein in the liver.	34, 35
PI*Q0 bellingham	rs199422211	VCV000017977	< 0.001%	The variant is a change from A to T in the DNA sequence of the *SERPINA1* gene. It results in no production of protein. It causes a **very severe plasma deficiency without accumulation** of protein in the liver.	36, 37, 38
PI*P lowell	rs121912714	VCV000017975	< 0.001%	The variant is a change from A to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is broken down more quickly. It causes a **severe plasma deficiency without accumulation** of protein in the liver.	39, 40, 41
PI*Q0 mattawa	rs763023697	VCV000552891	< 0.001%	The variant is an insertion of a T in the DNA sequence of the *SERPINA1* gene. It results in no production of protein. It causes a **very severe plasma deficiency without accumulation** of protein in the liver.	42, 43, 44
PI*Q0 clayton	rs764325655	VCV000188845	< 0.001%	The variant is an insertion of a C in the DNA sequence of the *SERPINA1* gene. It results in no production of protein. It causes a **very severe plasma deficiency without accumulation** of protein in the liver.	45, 46, 47

Variant tested	Associated genetic marker (rs)[1]	Clinical Significance of Variants - ClinVar accession number[2]	Global Minor Allele Frequency (MAF) %[3]	Biological explanation	References[4]
PI*M heerlen	rs199422209	VCV000017965	< 0.001%	The variant is a change from C to T in the DNA sequence of the *SERPINA1* gene. It results in a form of the protein that is broken down more quickly. It causes a **severe plasma deficiency without accumulation** of protein in the liver.	48, 49, 50

[1] https://www.ncbi.nlm.nih.gov/ (National Center for Biotechnology Information)

[2] https://www.ncbi.nlm.nih.gov/clinvar/ (ClinVar-National Center for Biotechnology Information)

[3] https://www.ncbi.nlm.nih.gov/snp/ (dbSNP-National Center for Biotechnology Information)

Minor Allele Frequency (MAF) in 1000 Genomes project. < 0.001% means that no frequency data are available in 1000 genomes project

[4] https://alphaidathome.com/references.pdf (References)

