Title: Alpha-1 Antitrypsin Deficiency *GeneReview* Table 5
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 5. Characteristics of Selected *SERPINA1* Alleles

PI Allele		Protein Level	Cellular Defect	Disease Association
Normal alleles	M (various)	Substitution (1 bp)	None	Normal
	$X_{christchurch}$	Glu363Lys	None	Normal
Deficiency alleles	S	Glu264Val	Intracellular degradation	Lung
	Z*	Glu342Lys	Intracellular accumulation	Lung, liver
	M_{malton}	Phe52del or p.Phe51del	Intracellular accumulation	Lung, liver
	S_{iiyama}	Ser53Phe	Intracellular accumulation	Lung
	$M_{heerlen}$	Pro369Leu	Intracellular degradation	Lung
	$M_{procida}$	Leu41Pro	Intracellular degradation	Lung
	$M_{mineral\ springs}$ *	Gly67Glu	Intracellular degradation	Lung
Null alleles	$QO_{granite\ falls}$	Tyr160Ter	No mRNA	Lung
	$QO_{ludwigshafen}$	Ile92Asn	No protein	Lung, liver
	$QO_{hongkong-1}$	Leu318LeufsTer17	Truncated; intracellular accumulation	Lung
	$QO_{isola\ di\ procida}$	17-kb del	Deletion of coding regions; No mRNA	Lung
Dysfunctional alleles	F	Arg223Cys	Defective neutrophil elastase inhibition	Lung
	Pittsburgh	Met358Arg	Antithrombin 3 activity	Bleeding diatheses
	$M_{mineral\ springs}$*	Gly67Glu	Defective neutrophil elastase inhibition	Lung
	Z*	Glu342Lys	Defective neutrophil elastase inhibition	Lung, liver

Adapted from DeMeo & Silverman [2004] with permission from the BMJ Publishing Group.

* Dysfunctional characteristics described are based on altered rates of association and inhibition of neutrophil elastase and deficiency characteristics.

References

DeMeo DL, Silverman EK. Alpha-1 antitrypsin deficiency, 2: genetic aspects of alpha(1)-antitrypsin deficiency: phenotypes and genetic modifiers of emphysema risk. Thorax. 2004;59:259–64.