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Antithrombin gene mutation 5356–5364*delCTT with type I deficiency and early-onset thrombophilia and a brief review of the antithrombin α -helix D molecular pathology

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In the original publication of this article, Tables 1 and 2 were unfortunately omitted. We apologise for this mistake. The correct versions of Tables 1 and 2 are given below.

Table 1 Antithrombin α -helix D amino acid residues with their major structural and functional impact. Mutation entries apply to human case studies only, whereas in vitro studies have generally not been considered

Residue	Structural/functional impact	Mutation	References
Ile 119	Helix packing, protein folding	delT in codon 119, frameshift, stop codon 126 type I deficiency	[20]
His 120	Helix packing, protein folding	His120Tyr, type I deficiency, misfolding delHis120–Phe121–Phe122, type I deficiency	[17, 19]
Phe 121–123	Helix packing, heparin binding, protein folding	delPhe, type I deficiency, misfolding	[5, 16, 24, this report]
Ala 124	Helix packing, protein folding		
Lys 125	Heparin binding		
Leu 126, Asn 127	Helix packing, protein folding	Leu126Pro, type I deficiency, misfolding	[25]
Cys 128	Disulfide bond formation with Cys 8 conformation of the heparin-binding site	Cys128Tyr, type I deficiency, misfolding, formation of high molecular weight AT species, impaired heparin-binding affinity	[7]
Arg 129	Heparin binding	Arg129Gln, AT Geneva, type II deficiency Arg129stop, type I deficiency	[8, 9, 16, 21, 24, 31]
Leu 130, Tyr 131	Helix packing, protein folding		
Arg 132, Lys 133	Interaction with shutter region, heparin binding	Arg132stop, type I deficiency	[25]
Ala 134	Linker between α -helix D and β -sheet A heparin binding		
Asn 135	Glycosylation site minor fraction unglycosylated (β -AT)	Asn135Thr, borderline AT deficiency, asymptomatic, glycosylation block, increased β -AT with high heparin affinity	[3]

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Table 2 Antithrombin (AT) gene mutation 5356–5364*delCTT leading to a deletion of a phenylalanine residue at codons 121–123. Summary of reported cases. *DVT* deep vein thrombosis, *PE* pulmonary embolism

Patient	AT activity (%)	AT antigen (%)	Comments	Reference
1	62	73		[5]
2	47	48	Thrombophlebitis	[16]
3	55	41	DVT/PE	[24]
4	38	43	Multiple DVT	This report