

Supplementary Table 1. Summary of C1-INH mutations in Chinese HAE patients. All published disease-causing mutations of the *SERPING1* gene are shown.

No.	Traditional genomic numbering	Nucleotide change	Amino acid change	Location	Refs
1	g.1812delT	c.44delT	p.L15Rfs78*	Exon2	[1]
2	g.2287_2288 delCA	c.143_144delCA	p.T48fs55*	Exon3	[2]
3	g.2350 delA	c.206delA	p.N69fs77*	Exon3	[2]
4	g.2433 C>T	c.289C>T	p.Q97*	Exon3	[1, 2]
5	g. 2439_2446del?	c.295_302delACCATCCAA	NA	Exon3	[3]
6	g.2440_2447 delCCATCCAA	c.296_303del CCATCCAA	p.T99fs129x	Exon3	[2]
7	g.2466 C>T	c.322C>T	p.Q108*	Exon3	[2]
8	g.2496 A>G	c.352A>G	p.T118A	Exon3	[2]
9	g.2577_2599del	c.433_455del	NA	Exon3	[3]
10	g.2605 A>G	c.461A>G	p.Y154C	Exon3	[2]
11	g.2653 C>T	c.509C>T	p.S170F	Exon3	[2]
12	g.2694 G>A	c.550G>A	p.G184R	Exon3	[2]
13	g.3864C<T	c.538C>T	p.Q180*	Exon3	[1]
14	g.4429_4445delTGAAGGGCTTCACGACC	c.629_645 delTGAAGGGCTTCACGACC	p.T210fs249x	Exon4	[2]
15	g.4485 + 1 G>A	c.685 + 1 G>A	Splicing defect	Intron4	[2]
16	g.8328 T>C	c.689T>C	p.L230P	Exon5	[2]
17	g.8334 T>A	c.695T>A	p.I232K	Exon5	[2]
18	g.9548_9549delCT	NA	NA	Exon5	[4, 5]
19	g.8366_8367 delCT	c.727_728delCT	p.T243fs254*	Exon5	[2]
20	g.9627_9628insCAC	NA	p.248_249insT	Exon5	[5]
21	g.9633_9635delCAA	NA	p.250delN	Exon5	[5]
22	g.8445_8447 insCAC	c.806_808insCAC	p.271insT	Exon5	[2]
23	g.8453_8455 delCAA	c.813_815delCAA	p.delN272	Exon5	[2]
24	g.8521 C>A	c.882C>A	p.Y294*	Exon5	[2]
25	g.9615G<A	c.794G>A	p.W265*	Exon5	[1]
26	g.8728 T>C	c.895T>C	p.W299R	Exon6	[2]
27	g.8729 G>A	c.896G>A	p.W299*	Exon6	[2]
28	g.9954delT	c.939delT	p.H314fs427*	Exon6	[1]
29	Intron6-1delG		Splicing defect	Intron6	[5]
30	g.14033 C>T	c.1042C>T	p.Q348*	Exon7	[2]
31	g.14112delG	c.1111delG	p.E371fs397*	Exon7	[2, 3]
32	g.14200_14201 insCC	c.1199_1200insCC	p.T404fs431*	Exon7	[2]
33	g.15397_15406delCCAGCCAGGA	c.1214_1223delCCAGCCAGGA	p.T405fs427*	Exon7	[1]
34	g.16671delC	c.1279delC	p.I426fs428*	Exon8	[2]
35	g.16681T>A	c.1289T>A	p.L430Q	Exon8	[2]
36	g.16714T>C	c.1326T>C	p.M441T	Exon8	[2]
37	g.16732 T>C	c.1340T>C	p.L447P	Exon8	[2, 3]
38	g.16734G>T	c.1342G>T	p.E448*	Exon8	[2]
39	g.16757delC	c.1365delC	p.S422fs433*	Exon8	[2]
40	NA	c.1391_1445del55	p.v464fsx556)	Exon8	[2]
41	g.16788 C>A	c.1396C>A	p.R466S	Exon8	[2]
42	g.16788 C>T	c.1396C>T	p.R466C	Exon8	[2, 6]
43	g.16789G>T	c.1397G>T	p.R466L	Exon8	[2]
44	g.16789G>A	c.1397G>A	p.R466H	Exon8	[3]
45	g.16810T>G	c.1418T>G	p.V473G	Exon8	[2]
46	g.16830G>A	c.1438G>A	p.V458M	Exon8	[3]
47	g.16871delG	c.1479delG	p.G493fs500*	Exon8	[2]
48	g.16872C>T	c.1480C>T	p.R472*	Exon8	[3]
49	g.16882A>G	c.1490A>G	p.D497G	Exon8	[2]
50	g.16884C>T	c.1492C>T	p.P476H	Exon8	[3]
51	g.17832delC	NA	p.fs408*	Exon8	[5]
52	g.17839delC	NA	p.fs408*	Exon8	[4]
53	g.17853delC	c.1279delC	p.L426fs428	Exon8	[1, 5]
54	g.17878C>A	NA	p.R444S	Exon8	[5]
55	g.18003T>C	NA	p.L425P	Exon8	[7]
56	g.18012G>A		p.V458M	Exon8	[5]

NA: not available; information was not provided in the original articles.

References

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