

**Table S1 Primers used for sequencing of *PROC* and *SERPINC1* gene**

|                 | Forward primer                 | Reverse primer              | Product size (bp) |
|-----------------|--------------------------------|-----------------------------|-------------------|
| <i>PROC</i>     |                                |                             |                   |
| Exon 1          | 5' GCTGGACGGCATCCTTGGT 3'      | 5' AAGGGACCTGAGACTGTGGCT 3' | 396               |
| Exon 2          | 5' GACAGCCCTTTTCATTCCG 3'      | 5' TTCCTGCCATTCCCCTCT 3'    | 316               |
| Exon 3          | 5' CGCCAAGCCATGACCTAG 3'       | 5' CATCCTAATCGCTCCACTCA 3'  | 458               |
| Exon 4-5        | 5' CCTAGCAGCCAACGACCATC 3'     | 5' GCCCACCTCCTCTAGGCAGTA 3' | 574               |
| Exon 6          | 5' GAGGGGGAGAGGTGGATG 3'       | 5' GCTCAACTCTGTTCTCGCTCC 3' | 360               |
| Exon 7          | 5' AGTGGTCTAAGTATCATTGGTT 3'   | 5' CAGGTTGCGTCCATCTTT 3'    | 442               |
| Exon 8          | 5' GCAAAGGCAGGAAGACACC 3'      | 5' CAGGGTGGGGGCACATAG 3'    | 473               |
| Exon 9          | 5' CTCCGTGACTCCTGAAAACC 3'     | 5' GGCTCCACCATGTAAACCTT 3'  | 1332              |
| <i>SERPINC1</i> |                                |                             |                   |
| Exon 1          | 5' GGTGTAACCATTTGTGTTTTTC 3'   | 5' GTCATTCCTGTGAGTCCTT 3'   | 639               |
| Exon 2          | 5' GGATTTAGCCTTTCTCTTGG 3'     | 5' GGTTGAGGAATCATTGGACT 3'  | 579               |
| Exon 3          | 5' CATCAGAACACAAGATTGAGCAT 3'  | 5' GCCCTCCAGCAGTCTTCA 3'    | 497               |
| Exon 4          | 5' TTAGATATGTGAGGCTTCCC 3'     | 5' ATTGACCTGCCCTGCTGA 3'    | 448               |
| Exon 5          | 5' CAATAACTATCCTCCTATGAATGT 3' | 5' TCGGGGTGGAGAAGGGAG 3'    | 484               |
| Exon 6          | 5' GTGAGAGTATGATTAGGTGAAGAT 3' | 5' GGAGGGAAGGCAGAACAA 3'    | 383               |
| Exon 7          | 5' GACCCAAATGTACTTTTTACTG 3'   | 5' TGTCTACCATCTTTTATCGC 3'  | 538               |

**Table S2 Summary for mutations of *SERPINC1* gene reported in Chinese**

| Mutation type | Nucleotide change   | Amino acid change        | AT:A level | AT:Ag level | Reference |
|---------------|---------------------|--------------------------|------------|-------------|-----------|
| Nonsense      | c.490C>T            | p.Arg164*                | Low        | Low         | [1]       |
| Nonsense      | c.211A>T            | p.Lys71*                 | Low        | Low         | [1]       |
| Nonsense      | c.6T>A              | p.Tyr2*                  | Low        | Low         | [1]       |
| Nonsense      | c.229_236delinsGGGA | p.Asn77Glyfs*35          | Low        | Low         | [1]       |
| Splicing      | c.41+2T>C           | -                        | Low        | Low         | [1]       |
| Deletion      | c.1066_1083del      | p.Arg356_Phe361del       | Low        | Low         | [1]       |
| Missense      | c.641C>A            | p.Ser214Tyr              | Low        | Low         | [1]       |
| Deletion      | c.1066_1083del      | p.Arg356_Phe361del       | Low        | Low         | [1]       |
| Nonsense      | c.116_123del        | p.Ile39Serfs*25          | Low        | Low         | [1]       |
| Nonsense      | c.484_485del        | p.Tyr163Serfs*25         | Low        | Low         | [1]       |
| Deletion      | c.1030_1032del      | p.Glu344del              | Low        | Low         | [1]       |
| Nonsense      | c.1016G>A           | p.Trp339*                | Low        | Low         | [1]       |
| Missense      | c.719A>G            | p.Asn240Ser              | Low        | Low         | [1]       |
| Nonsense      | c.849_853dupGATGT   | p.Met284Metfs*1          | Low        | Low         | [1]       |
| Missense      | c.458T>A            | p.Phe153Tyr              | Normal     | Normal      | [1]       |
| Missense      | c.491G>A            | p.Arg164Gln              | Normal     | Normal      | [1]       |
| Missense      | c.598G>C            | p.Ala200Pro              | Normal     | Normal      | [1]       |
| Missense      | c.883G>A            | p.Val295Met              | Normal     | Normal      | [1]       |
| Missense      | c.1277C>T           | p.Ser426Leu              | Normal     | Normal      | [1]       |
| Deletion      | c.86_87delinsCT     | p.Cys29Ser               | Normal     | Normal      | [1]       |
| Missense      | c.880C>T            | p.Arg294Cys              | Normal     | Normal      | [1]       |
| Missense      | c.883G>A            | p.Val295Me               | Normal     | Normal      | [1]       |
| Missense      | c.938T>C            | p.Met313Thr              | Normal     | Normal      | [1]       |
| Missense      | c.389C>T            | p.Thr130 (98) Ile        | Low        | Low         | [2]       |
| Missense      | c.464T>G            | p.Phe155 (123) Cys       | Low        | Low         | [2]       |
| Missense      | c.737T >A           | p.Val246 (214) Glu       | Low        | Low         | [2]       |
| Missense      | c.817A>G            | p.Lys273 (241) Glu       | Low        | Normal      | [2]       |
| Missense      | c.1114C>T           | p.Leu372 (340) Phe       | Low        | Low         | [2]       |
| Missense      | c.1202A>G           | p.His401 (369) Arg       | Low        | Low         | [2]       |
| Missense      | c.1274G>A           | p.Arg425 (393) His       | Low        | Normal      | [2]       |
| Nonsense      | c.6T>G              | p.Tyr2 (-31) *           | Low        | Low         | [2]       |
| Nonsense      | c.203C>G            | p.Ser68 (36) *           | Low        | Low         | [2]       |
| Nonsense      | c.304A>T            | p.Lys102 (70) *          | Low        | Low         | [2]       |
| Nonsense      | c.1016G>A           | p.Try339 (307) *         | Low        | Low         | [2]       |
| Nonsense      | c.1171C>T           | p.Arg391 (359) *         | Low        | Low         | [2]       |
| Nonsense      | c.1185T>A           | p.Tyr395 (363) *         | Low        | Low         | [2]       |
| Nonsense      | c.337_338delCT      | p.Pro112 (80) fs*14      | Low        | Low         | [2]       |
| Deletion      | c.460_462delTTC     | p.Phe154 (122) del       | Low        | Low         | [2]       |
| Missense      | g.7920C>T           | p.Trp225Cys              | Low        | Low         | [3]       |
| Missense      | g.13863C>A          | p.Ala404Asp              | Low        | Low         | [3; 4]    |
| Compound      | c.134G>A & c.342T>G | p.Arg45Gln & p.Ser114Arg | Low        | Low         | [4; 5; 6] |

|          |                      |                    |        |         |         |
|----------|----------------------|--------------------|--------|---------|---------|
| Deletion | c.337-338del         | p.Leu113fs         | Low    | Unknown | [7]     |
| Nonsense | c.770G>A             | p.Trp257*          | Low    | Low     | [4; 7]  |
| Deletion | c.800-803del         | p.Arg267fs         | Low    | Normal  | [4; 7]  |
| Missense | g.1267G >A (p.A391T) | p.Ala391Thr        | Normal | Normal  | [8]     |
| Missense | g. 8263 C>T          | Leu340Phe          | Low    | Low     | [9]     |
| Deletion | g. 5894- 6 del TTC   | Phe122del          | Low    | Low     | [8]     |
| Missense | g. 5898 T>G          | Phe123Cys          | Low    | Low     | [8]     |
| Nonsense | g:3239-3240delCT     | Pro80Profs14*      | Low    | Low     | [10]    |
| Nonsense | g:3206A>T            | p.Lys70*           | Low    | Low     | [9]     |
| Missense | g:13328G>A           | p.Ala404Thr        | Low    | Low     | [11]    |
| Missense | g:2757C>T            | p.Thr98Ile         | Low    | Low     | [7; 12] |
| Nonsense | g:13389delG          | p.Arg425Glufs*1    | Low    | Low     | [13]    |
| Nonsense | g:10381delT          | p.Phe400Serfs*5    | Low    | Low     | [14]    |
| Missense | g:13830G>A           | p.Arg393His        | Low    | Normal  | [15]    |
| Nonsense | g:610G>T             | p.Tyr31*           | Low    | Low     | [16]    |
| Nonsense | g:18390-1insCT       | p.Thr401*          | Low    | Normal  | [17]    |
| Nonsense | g:10446C>T           | p.Arg142*          | Low    | Low     | [17]    |
| Missense | g:7747T>C            | p.Leu78Pro         | Low    | Normal  | [17]    |
| Missense | g:7386G>C            | p.Trp255Cys        | Low    | Low     | [18]    |
| Nonsense | g:2591C>T            | p.Ser36*           | Low    | Low     | [18]    |
| Nonsense | g:9819C>T            | p.Arg359*          | Low    | Low     | [18]    |
| Nonsense | g:9833T>A            | p.Tyr363*          | Low    | Low     | [19]    |
| Deletion | c.1061_1078del       | p.Phe355_Gly360del | Low    | Low     | [20]    |
| Nonsense | c.1033_1035 del      | p.Glu345del        | Low    | Low     | [21]    |
| Missense | c.442T>C             | p.Ser148Pro        | Low    | Low     | [21]    |
| Missense | c.235C>T             | p.Arg79Cys         | Low    | Low     | [21]    |

AT:A: antithrombin activity; AT:Ag: antithrombin antigen

**Table S3 Summary for mutations of *PROC* gene reported in Chinese**

| Type     | Nucleotide change                              | Amino acid change                                             | PC:A level | PC:Ag level | Ref.     |
|----------|------------------------------------------------|---------------------------------------------------------------|------------|-------------|----------|
| Missense | g.6219G>A                                      | p.Arg169Gln                                                   | Low        | Low         | [22; 23] |
| Missense | g.6128T>G                                      | p.Phe139Val                                                   | Low        | Low         | [24]     |
| Missense | g.8478G>C                                      | p.Asp255His                                                   | Low        | Low         | [24]     |
| Compound | g.26T>C & g.8599C>T                            | p.Leu8(−34)Pro &<br>p.Thr337 (295)Ile                         | Low        | Low         | [25]     |
| Compound | g.1474G>A & g.6152C>T                          | p.Glu71(29)Lys &<br>p.Arg189 (147)Trp                         | Low        | Low         | [25]     |
| Compound | g.6245C>T & g.8478G>C                          | p.Arg220 (178)Trp &<br>p.Asp297 (255)His                      | Low        | Low         | [25]     |
| Compound | g.5498C>T & g.6152C>T                          | p.Arg57 (15) Trp &<br>p.Arg189 (147)Trp                       | Low        | Low         | [25]     |
| Compound | g.26T>C &<br>g.7212 C>T &<br>g.6161-6163delAAG | p.Leu8 (−34)Pro &<br>p.Ala251 (209)Val &<br>p.Lys192 (150)del | Low        | Low         | [25]     |
| Compound | g.6152C>T<br>c.1680T>Ac                        | p.Arg189 (147)Trp &<br>p.Tyr560X                              | Low        | Low         | [25]     |
| Compound | g.6152C>T<br>g.6245C>T                         | p.Arg189 (147)Trp &<br>p.Arg220(178)Trp                       | Low        | Low         | [25]     |
| Missense | g.5498 C>T                                     | p.Arg57(15)Trp                                                | Low        | Low         | [25]     |
| Missense | g.3426G>T                                      | p.Asp170(128)Tyr                                              | Low        | Normal      | [25]     |
| Missense | g.3442G>A                                      | p.Cys175(133)Tyr                                              | Low        | Low         | [25]     |
| Missense | g.6152C>T                                      | p.Arg189(147)Trp                                              | Low        | Low         | [25]     |
| Deletion | g.6161-6163delAAG                              | p.Lys192(150)del                                              | Low        | Low         | [25]     |
| Missense | g.8478G>C                                      | p.Asp297(255)His                                              | Low        | Unknown     | [25]     |
| Missense | g.8746T>C                                      | p.Leu386(344)Pro                                              | Low        | Normal      | [25]     |
| Missense | g.8763G>A                                      | p.Gly392(350)Arg                                              | Low        | Normal      | [25]     |
| Missense | g.8807G>A                                      | p.Met406(364)Ile                                              | Low        | Normal      | [25]     |
| Nonsense | g.8831G>A                                      | p.Trp414(372)X                                                | Low        | Low         | [25; 26] |
| Missense | Unknown                                        | p.Thr315Asn                                                   | Low        | Low         | [27]     |
| Missense | g.5498C>T                                      | p.Arg15Trp                                                    | Low        | Low         | [25; 28] |
| Missense | c.889G>C                                       | p.Asp297His                                                   | Low        | Normal      | [29]     |
| Missense | c.1258G>T                                      | p.Val420Leu                                                   | Low        | Normal      | [29]     |
| Compound | c.889G>C & c.1258G>T                           | p.Asp297His & p.Val420Leu                                     | Low        | Normal      | [29]     |
| Missense | g.6128T>G                                      | p.Phe139Val                                                   | Low        | Low         | [30]     |
| Missense | g.8478G>C                                      | p.Asp255His                                                   | Low        | Low         | [30]     |
| Compound | g.6128T>G & g.8478G>C                          | p.Phe139Val & Asp255His                                       | Low        | Low         | [30]     |
| Missense | c.842A>T                                       | p.Asp281Val                                                   | Low        | Unknown     | [31]     |
| Compound | c.541T>G & c.565C>T                            | p.Phe181Val & p.Arg189Trp                                     | Low        | Unknown     | [31]     |
| Splicing | c.400+1G>C                                     | -                                                             | Low        | Unknown     | [31]     |
| Deletion | c.574_576del                                   | p.Lys192del or p.Lys193del                                    | Low        | Normal      | [31]     |
| Compound | g.5540G>A & g.10230C>T                         | p.Glu29Lys & p.Arg147Trp                                      | Low        | Low         | [32]     |
| Missense | g.5540G>A                                      | p.Glu29Lys                                                    | Low        | Low         | [32]     |

|          |                                         |                                         |        |         |             |
|----------|-----------------------------------------|-----------------------------------------|--------|---------|-------------|
| Missense | g:10230C>T                              | p.Arg147Trp                             | Low    | Low     | [32; 33]    |
| Missense | g:3135 c>G                              | p.Cys64Trp (PC Shanghai)                | Low    | Low     | [34]        |
| Missense | g:6128T>G                               | p.Phe139Val                             | Low    | Low     | [34; 35]    |
| Compound | g:3135 c>G & g:6128T>G                  | p.Cys64Trp & p.Phe139Val                | Low    | Low     | [34]        |
| Deletion | g:1616-1613del                          | p.Lys150del                             | Normal | Normal  | [34]        |
| Deletion | g:6164-6166del                          | p.Lys151del                             | Normal | Normal  | [34]        |
| Missense | c.508G>T                                | p.Asp170Tyr                             | Low    | Unknown | [4; 36]     |
| Deletion | c.577-579del                            | p.Lys192del                             | Low    | Unknown | [4; 36]     |
| Missense | c.1174G>A                               | p.Gly392Arg                             | Low    | Unknown | [4; 36]     |
| Missense | c.1157T>C                               | p.Leu386Pro                             | Low    | Unknown | [4; 36]     |
| Missense | c.524G>A                                | p.Cys175Tyr                             | Low    | Unknown | [4; 36]     |
| Missense | c.856G>A                                | p.Gly266Arg                             | Low    | Unknown | [4]         |
| Compound | c.541T>G & c.595C>T                     | p.Phe181Val & p. Arg199*                | Low    | Unknown | [4]         |
| Nonsense | c.740G>A                                | p.Trp247*                               | Low    | Unknown | [4; 37]     |
| Missense | c.1218G>A                               | p.Met406Ile                             | Low    | Unknown | [4; 25]     |
| Missense | c.752C>T                                | p.Ala251Val                             | Low    | Unknown | [4]         |
| Missense | c.565C>T                                | p.Arg189Trp                             | Low    | Unknown | [4; 36; 38] |
| Missense | c.949C > T                              | p.Pro275Ser                             | Low    | Low     | [39]        |
| Compound | 6245C>T & 8478G>C                       | p.Arg178Trp & p. Asp255His              | Low    | Low     | [40]        |
| Compound | 26T>C & 8599C>T                         | p.Leu34Pro & p.Thr295Ile                | Low    | Low     | [40]        |
| Compound | c.889G. & c.1258G>T                     | p.Asp297His & p.Val420Leu               | Low    | Low     | [38]        |
| Splicing | c.*73C>T                                | -                                       | Low    | Low     | [38]        |
| Missense | c.524G>A                                | p.Cys175Tyr                             | Low    | Low     | [38]        |
| Nonsense | c.716dupG                               | p.Ala240GlyfsX17                        | Low    | Low     | [38]        |
| Missense | c.632G>A                                | p.Arg211Gln                             | Low    | Low     | [38]        |
| Compound | c.349_352del & c.541T.G                 | p.Phe118AlafsX16& p.Phe181Va            | Low    | Low     | [38]        |
| Splicing | c.400+5G>A                              | -                                       | Low    | Low     | [38]        |
| Missense | c.316T>G                                | p.Cys106Gly                             | Low    | Low     | [38]        |
| Missense | c.889G>C & c.891C>T                     | p.Asp297His                             | Low    | Low     | [38]        |
| Missense | c.935C>T                                | p.Ser312Leu                             | Low    | Low     | [38]        |
| Missense | c.658C>T                                | p.Arg220Trp                             | Low    | Low     | [38]        |
| Compound | c.541T>G & c.980A>T                     | p.Phe181Val p.Glu327Val                 | Low    | Low     | [38]        |
| Missense | c.208A>G                                | p.Lys70Glu                              | Low    | Low     | [38]        |
| Splicing | c.237+5G>A                              | -                                       | Low    | Low     | [38]        |
| Missense | c.669C>A                                | p.Ser223Arg                             | Low    | Low     | [38]        |
| Missense | c.541T>G                                | p.Phe181Val                             | Low    | Low     | [38]        |
| Missense | g:1265C>T                               | p:Pro275Ser                             | Low    | Low     | [41]        |
| Compound | c.595C>T & c.541 T>G                    | p.Phe181Val & p.Arg199*                 | Low    | Low     | [37]        |
| Compound | nt26T>C & nt7212C>T & nt6166-6163delAAG | p:Leu34Pro & p. Ala209Val & p.Lys150del | Low    | Low     | [42]        |
| Missense | c.1069A>G                               | p.Thr357Ala                             | Low    | Low     | [21]        |
| Missense | c.970G>A                                | p.Gly324Ser                             | Low    | Low     | [21]        |
| Missense | c.541T>G                                | p.Phe181Val                             | Low    | Low     | [21]        |
| Missense | c.658C>T                                | p.Arg220Trp                             | Low    | Low     | [21]        |

|          |              |             |     |     |      |
|----------|--------------|-------------|-----|-----|------|
| Deletion | c.577_579del | p.Lys193del | Low | Low | [21] |
| Missense | c.1019C>T    | p.Thr340Met | Low | Low | [21] |
| Missense | c.247T>C     | p.Trp83Arg  | Low | Low | [21] |
| Missense | c.487G>A     | p.Ala163Thr | Low | Low | [21] |
| Missense | c.865C>T     | p.Pro289Ser | Low | Low | [21] |
| Missense | c.262G>A     | p.Asp88Asn  | Low | Low | [21] |

PC:A: protein C activity; PC:Ag: protein C antigen

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