**Supplemental Table**

Characteristics of the genetic defects and onset of thrombotic episodes in the 15 unrelated Chinese subjects carrying Arg147Trp and Lys150del variants

| patient No. | Age/ sex | Activity (%) | Antigen (%) | Nucleotide variation (DNA) | Amino acid substitution | Thrombotic episodes (onset age) | Thrombophilic factors | Thrombotic episodes (family member) |
|---|---|---|---|---|---|---|---|---|
| 1 | 20/M | 19.0 | 42.0 | g.1474G>A<br>g.6152C>T | p.Glu29 Lys<br>p.Arg147Trp | PE (19) | | DVT, PVT (father with p.Glu29Lys) |
| 2 | 27/M | 39.0 | 42.1 | g.5498C>T<br>g.6152C>T | p.Arg15Trp<br>p.Arg147Trp | DVT(25), DVT(26) | | |
| 3 | 24/M | 46.0<br>19.5[a] | 64.4<br>20.9[b] | g.6152C>T<br>c.1680T>A[c] | p.Arg147Trp<br>P.Tyr519X[d] | PE (22), DVT(22), DVT (24) | Protein S deficiency | DVT (father with p.Tyr519X in PROS) |
| 4 | 47/M | 30.0 | 48.4 | g.6152C>T<br>g.6245C>T | p.Arg147Trp#<br>p.Arg178Trp# | DVT (40y), DVT (43) | Cholecystectomy | DVT (brother with the double mutations) |
| 5 | 24/F | 38.6 | 45.3 | g.6152C>T | p.Arg147Trp | DVT (23), DVT (24) | | |
| 6 | 68/M | 46.8 | 67.2 | g.6152C>T | p.Arg147Trp | Both DVT and PE (68) | Cholecystectomy | |
| 7 | 24/F | 65.9 | 76.5 | g.6152C>T | p.Arg147Trp | DVT (21) | | |
| 8 | 33/F | 63.9 | 71.8 | g.6152C>T | p.Arg147Trp | DVT (33) | Puerperium | |
| 9 | 19/M | 69.4 | 74.9 | g.6152C>T | p.Arg147Trp | DVT (19) | | |
| 10 | 28/M | 63.9 | 64 | g.6152C>T | p.Arg147Trp | DVT (28) | | |
| 11 | 32/F | 1.2 | 0 | c.3135C>G<br>c.6128T>G | p.Cys64Trp<br>p.Phe139Val | DVT (24), MVT (30), DVT(31) | | DVT, MVT and disseminated thrombosis (mother with p.Cys64Trp) |
| | 6/F* | 30.5 | 54.8 | c.6128T>G<br>c.6161-6163delAAG | p.Phe139Val<br>p.Lys150del | | | |
| 12 | 30/M | 36.0 | 57.0 | c.26T>C<br>c.7212 C>T<br>c.6161-6163delAAG | p.Leu-34Pro#<br>p.Ala209Val#<br>p.Lys150del | DVT (28y), DVT (30) | Protein S deficiency | DVT (mother with p.Lys150del combined with protein S deficiency ), MVT (brother with the double mutations) |
| | proteinS | 18.0[a] | 23.7.[b] | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| 13 | 19/M | 50.3 | 61.3 | c.6161-6163delAAG | p.Lys150del | Left CVT (19) DVT (19) |
| 14 | 28/M | 68.7 | 68.2 | c.6161-6163delAAG | p.Lys150del | DVT (27) |
| 15 | 18/F | 56.0 | 44.0 | c.6161-6163delAAG | p.Lys150del | |

PC:A: 70-140%; PC:Ag: 70-130%; FPS:A: 63~135%; FPS: Ag: 60~150%.

a: FPS:A; b: FPS: Ag; c: PS cDNA; d: PS amino acid substitution; #: double mutations: *daughter of the Patient 11

DVT: deep venous thrombosis (indicating the low extremity here); PE: pulmonary embolism; PVT: portal venous thrombosis; MVT: mesenteric venous thrombosis; AVT: axillary vein thrombosis; ND: not detected

Clinical data were obtained as described by Ding et al in Ref. 16.