

Incidence and features of thrombosis in children with inherited antithrombin deficiency

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| Case | Sex | Age of Thrombosis (years old) | Thrombosis | Risk Factors | Other Thrombophilias | Recurrence/Death/ Severe Complications | Mutation | Anti-Fxa (%) | Ag AT (%) | AT Def. | Cohort |
|------|-----|-------------------------------|--------------------------------|--------------------------------|---|--|----------------------------|--------------|-----------|----------|--------|
| 1 | M | neonate | Left arm thrombosis | | | Arm amputation | p.Arg425His | 54 | 93 | II (RS) | 1 |
| 2 | M | neonate | CSVT | Forceps | | Psychomotor delay | p.Met283Val | 39 | 71 | II (RS) | 1 |
| 3 | F | neonate | Aortic thrombosis | | | Deceased | p.Leu131Phe (homozygosis) | 11 | nd | II (HBS) | 1 |
| 4 | M | neonate | CSVT | Infection | | | p.Arg161* | 52 | 52 | I | 1 |
| 5 | M | neonate | Massive thrombosis | Sepsis | | Deceased | p.Pro112Ser | nd | nd | I | 1 |
| 6 | M | neonate | DVT | | | Deceased | p.Leu131Phe (homozygosis) | 34* | nd | II (HBS) | 2 |
| 7 | M | neonate | CSVT | | | | p.Gln150Pro | 55 | nd | II (HBS) | 2 |
| 8 | F | neonate | myocardial infarction and CSVT | Clavicle fracture | | Recurrences | p.Gly456Arg | 48 | 51 | I | 2 |
| 9 | M | neonate | CSVT | | | | p.Met284Lys | 43 | 39 | I | 2 |
| 10 | M | neonate | DVT and CSVT | | Heterozygous Factor V Leiden | | p.Phe94Ile fs X 19 | 50 | 46 | I | 2 |
| 11 | F | neonate | DVT and CSVT | Vacuum extraction | | Tetraplegic | p.Ile218Asn | 68 | nd | nd | 2 |
| 12 | F | neonate | DVT and arterial thrombosis | Cardiac surgery | | | p.Arg391* | 45 | 48 | I | 2 |
| 13 | M | neonate | CSVT and renal thrombosis | | | | p.Arg391Profs*3 | nd | nd | I | 1 |
| 14 | M | neonate | renal VT and DVT | Prematurity, fetal distress | | | p.Phe376Cys | 43 | nd | nd | 2 |
| 15 | M | 1 | CSVT and DVT | | Heterozygous Factor V Leiden | | p.Ile115Asn | 65 | 50 | I | 1 |
| 16 | M | 2 | CSVT | | | Deceased | p.Val137Ala | 45 | nd | I | 1 |
| 17 | F | 2 | CSVT | Cesarean section | | | p.Thr243Phe fs*40 | 46 | nd | I | 2 |
| 18 | F | 5 | CSVT | | | | p.Arg161* | 59 | 57 | I | 1 |
| 19 | M | 5 | CSVT | Infection | | | c.762 +1 G>A | 59 | nd | nd | 2 |
| 20 | F | 6 | DVT | | | | p.Met370Thr | 78 | nd | I | 1 |
| 21 | M | 6 | Arterial thrombosis | | | | p.Met370Thr | 78 | nd | I | 1 |
| 22 | M | 8 | DVT | Trauma | | Recurrence (15 y) | p.Cys53Stop | 49 | nd | I | 2 |
| 23 | M | 9 | DVT | | | Recurrence | c.1154-14 G>A | 47 | 44 | I | 2 |
| 24 | M | 11 | DVT | | | Recurrences (11, 12, 17y) | PM2-CDG | 67 | 60 | I | 1 |
| 25 | M | 12 | DVT | Immobilisation of legs | | | p.Ser223Pro | 49 | 43 | I | 1 |
| 26 | M | 12 | PE and DVT | | | | p.Arg445Serfs*17 | 37 | 42 | I | 1 |
| 27 | F | 12 | DVT and PE | Surgery | | Recurrence | p.Trp257Arg | 59 | nd | I | 2 |
| 28 | F | 13 | DVT | Oral contraceptives | | Recurrences | deletion exon 5 | 41 | 41 | I | 2 |
| 29 | M | 13 | PE and DVT | | | | Complete SERPINC1 deletion | 52 | 46 | I | 1 |
| 30 | F | 13 | DVT | | | | p.Arg161* | nd | nd | I | 1 |
| 31 | F | 13 | DVT | | | | p.Leu131Phe (homozygosis) | nd | nd | II (HBS) | 2 |
| 32 | M | 14 | PE | | | | p.Ser381Pro | 56 | 66 | I | 1 |
| 33 | F | 14 | Renal VT | | | Recurrence, deceased | p.Met11le | 55 | 57 | I | 1 |
| 34 | M | 14 | DVT | Renal agenesis | | Recurrence (16y) | p.Pro318Leu | 55 | 88 | II (RS) | 2 |
| 35 | M | 15 | DVT | Long flight | | Recurrences (27,42y) | Intron 3 c.625-2A>G | 44 | 47 | I | 1 |
| 36 | M | 15 | DVT | May Thurner Syndrome | Heterozygous Factor V Leiden | | p.Pro112Leu | 40 | 40 | I | 1 |
| 37 | M | 15 | DVT | | | | p.Arg229* | 56 | nd | I | 1 |
| 38 | M | 15 | PE | Infection: varicella | | | p.Lys157Arg | 60 | 60 | I | 1 |
| 39 | F | 15 | DVT and PE | Oral contraceptives | | | p.Pro384Pro | 58 | nd | I | 2 |
| 40 | F | 15 | Extensive PE | Oral contraceptives | | | p.Thr185Pro fs*99 | 46 | nd | I | 2 |
| 41 | F | 15 | DVT | Oral contraceptives | | | p.Pro318Leu | 65 | 64 | II (RS) | 2 |
| 42 | M | 15 | DVT | | | | p.Ser223Pro | 52 | 39 | I | 1 |
| 43 | F | 16 | DVT and PE | | | | c.1154-14 G>A | 40 | 57 | I | 1 |
| 44 | F | 16 | DVT | Puerperium | | Recurrence | p.Pro112Ser | 60 | nd | I | 1 |
| 45 | M | 16 | DVT and PE | | | | not found | 66 | 57 | I | 1 |
| 46 | F | 16 | DVT | | Heterozygous Factor V Leiden | | p.Leu131Phe | 53 | nd | II (HBS) | 2 |
| 47 | F | 16 | PE | | | | Complete SERPINC1 deletion | 55 | nd | I | 2 |
| 48 | M | 16 | DVT | Trauma | | | p.Leu131Phe (homozygosis) | nd | nd | II (HBS) | 2 |
| 49 | M | 17 | PE | | | | p.Ser114Asn | 48 | 47 | I | 1 |
| 50 | F | 17 | DVT | Surgery | | Recurrence | p.Met370Thr | 64 | nd | I | 1 |
| 51 | F | 17 | DVT | Puerperium | | Recurrence | p.Pro112Ser | 54 | nd | I | 1 |
| 52 | F | 17 | DVT | Pregnancy | | Recurrence | p.Pro112Ser | 60 | nd | I | 1 |
| 53 | M | 17 | DVT | | | | duplication exon 6 | 41 | 53 | I | 1 |
| 54 | M | 17 | DVT | | | | p.Arg161* | 61 | 50 | I | 1 |
| 55 | F | 17 | DVT | | | | p.Arg425del | 59 | 100 | II (RS) | 1 |
| 56 | F | 17 | DVT and PE | Oral Contraceptives and trauma | | | p.Met284Thr | 37 | 38 | I | 2 |
| 57 | F | 17 | DVT and PE | | | Recurrences (29, 36y) | p.Arg391* | 59 | 53 | I | 2 |
| 58 | F | 17 | DVT and PE | Oral contraceptives | | | p.Asn224Glu fs X 55 | 42 | nd | I | 2 |
| 59 | M | 17 | PE | Trauma | | Recurrence (18 y) | p.Thr185Pro fs*99 | 52 | 45 | I | 2 |
| 60 | F | 17 | DVT and PE | Oral contraceptives | | | c.1154-14 G>A | 61 | 46 | I | 2 |
| 61 | M | 17 | DVT | | | Recurrence (26 y) | p.Gln70Asp fs*33 | 53 | 62 | I | 2 |
| 62 | F | 18 | CSVT | Oral contraceptives | Heterozygous prothrombin G20210A mutation | | Lys165Glu fs*7 | 44 | 43 | I | 1 |
| 63 | M | 18 | DVT | Infection | | | p.Gly456Arg | 50 | 60 | I | 1 |
| 64 | F | 18 | PE | Oral contraceptives | | Deceased | p.Ser426Leu | 60 | nd | II (RS) | 1 |
| 65 | M | 18 | DVT and PE | | | | p.Leu162His | 54 | nd | I | 2 |
| 66 | F | 18 | DVT | Oral contraceptives | | | c.1154-14 G>A | 64 | 72 | I | 2 |
| 67 | M | 18 | DVT | Plastering of leg | | Recurrence (22y) | not found | 57 | 48 | I | 2 |
| 68 | M | 18 | DVT | Obesity(105Kg) surgery | Homozygous Factor V Leiden | | p.Leu131Phe | 52 | 78 | II (HBS) | 2 |
| 69 | F | 18 | DVT | Oral contraceptives | | | p.Leu372Pro | 44 | 48 | I | 2 |
| 70 | F | 18 | DVT | Oral contraceptives | | Recurrence (18y) | p.Arg229* | 53 | 50 | I | 2 |
| 71 | F | 18 | DVT | | | | p.Arg79His | 65 | nd | II (HBS) | 2 |
| 72 | M | 18 | DVT | | | | c.1154-2 A>G | 50 | 54 | I | 2 |
| 73 | M | 18 | PE and DVT | | | | p.Arg445Serfs*17 | 31 | 40 | I | 1 |

Abbreviations:CSVT (Cerebral sinovenous thrombosis); MTHFR (methylentetrahydrofolate reductase); DVT (Deep vein thrombosis); PE (Pulmonary embolism); HBS (Heparin binding site); RS (Reactive site). Nd: not determined
* patient number 6: anti-Fxa measured 4 days after birth and after plasma transfusion.