Background Prothrombotic risk factors (PRF) are suggested to be involved in the pathogenesis of PAIS. However, most of published studies are retrospective, they vary in the PRF tested and parents are not usually investigated.

Objective To determine the impact of parental and infant thrombophilia on neonatal cases diagnosed of PAIS in a prospective case-control study.

Methods Factor V (G1691A mutation), prothrombin G20210A variant, MTHFR C677T genotype, antithrombin, protein C, protein S, lipoprotein (a), homocystein (Hcy) and anticardiolipin antibodies were investigated in 45 infant-parent pairs with PAIS and in 85 controls. Blood samples were drawn within the first week of life.

Results All thrombophilic factors investigated were similar or even less frequent among patients with PAIS and their parents compared to controls. The most frequent PRFs were Hcy > 11 and MTHFR homozygosity in cases (7.5% and 5.3%, respectively) and in controls (24.4% and 13.1%, respectively). Thirteen neonates diagnosed of PAIS (28.9%) had at least 1 PFR, compared to 39 subjects (44.3%) in the control group (OR/95% CI, 0.51/0.23 to 1.10) (p = 0.001). Twenty three mothers of infants with PAIS (51.1%) were positive for thrombophilia markers, compared to 49 (55.7%) controls (p = .617). In 8 mother-infant pairs (17.8%), at least 1 PFR could be identified for either mother or infant, compared to 19 controls (21.6%). Fifteen neonates with stroke (33.3%) had at least one PFR compared to the 57 subjects (64.8%) in the control group (p < 0.001).

Conclusion Our data do not support that PFR play a major role in PAIS.