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Letter to the Editor

Prevalence of genetic thrombophilia in primary antiphospholipid syndrome

Dear Editor,

Primary antiphospholipid syndrome (pAPS) is characterized by thrombotic events and pregnancy losses associated with the presence of antiphospholipid antibodies. The combination of two or more thrombophilic factors increases the risk of thrombosis[1-5]. The present study aimed to assess the prevalence of other genetic or acquired thrombophilic factors in pAPS and verify whether there is a relevant clinical association. We investigated a cohort of 62 patients (88.9% women; mean age: 39.3 years) diagnosed with pAPS (Sapporo criteria), the presence of other thrombophilic factors, namely: hyperhomocysteinemia (HH) by chemiluminescence method; protein S deficiency (DPS) through functional quantification by chronometric method; protein C (DPC) and antithrombin III (ATIII) deficiencies through functional quantification by the chromogenic substrate; and the search for the G20210A mutation of the prothrombin gene (MGP) by polymerase chain reaction (PCR), followed by digestion with Hind III restriction enzyme and the search for the Q506 mutation of the factor V gene (Leiden factor V - LFV), by PCR followed by digestion with restriction enzyme Mnl I.

Three (19%) patients with LFVL in heterozygous form were found in 16 surveyed. HH was found in 2/40 (5%); 1/31 FVDATIII (3.2%); DPC at 7/33 (21%); and DPS at 8/32 (32%). No MGP was found in the 14 patients in which it was searched. It was also found that when comparing the groups of isolated APS with those of combined APS with thrombophilia factors, it was found that the age of the first presentation of thrombosis or pregnancy loss was significantly lower in the isolated APS group (30.4 *vs*. 39 years, $p=0.018$). On the other hand, there were no significant differences between the frequency of arterial events (ischemia of the extremities, CVS), venous (deep venous thrombosis, pulmonary embolism), and obstetric events between patients with isolated and combined APS. The groups also did not differ about the frequency and anticardiolipin IgG, IgM, and lupus anticoagulant ($p>0.05$).

In conclusion, patients with APS may have other thrombophilia in up to a third of cases, mainly due to deficiency of anticoagulant proteins, and the thrombotic or obstetric clinical event appears later than in the group of patients with APS with other thrombophilia.

Conflict of Interest

The Authors declare that they have no conflict of interests.

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