Molecular thrombophilic profile in Mexican patients with idiopathic recurrent pregnancy loss

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ABSTRACT. Idiopathic recurrent pregnancy loss (IRPL) is defined by three or more consecutive miscarriages occurring before the twentieth week of gestation as a result of unidentified etiological factors. The results of previous studies have indicated that prothrombotic factors play a pathogenic role in early and late pregnancy. This study aimed to identify inherited prothrombotic and hypofibrinolytic risk factors

in Mexican-Mestizo patients with IRPL. Fifty-six women with IRPL and 50 control women with at least two full-term pregnancies and no history of RPL were included in this case-control study. Four prothrombotic (F5 G1691A, F2 G20210A, MTHFR C677T-A1298C) and one hypofibrinolytic (PAI1 4G/5G) restricted fragment length polymorphisms were subjected to molecular analysis. In the case of hypofibrinolytic ACE Ins/Del (I/D), identification was performed by direct PCR. The independent risk correlated with the presence of polymorphisms in IRPL patients was estimated using odds ratio (OR) with a 95% confidence interval (CI). *MTHFR* 677TT was the most frequent prothrombotic factor in the IRPL group (23%), followed by the compound-heterozygous *C677T*-A1298C (16%) and heterozygous *F2* 20210GA (3.6%). The heterozygous *ACE* I/D (62%) was the main hypofibrinolytic risk factor of IRPL, followed by the homozygote *PAI1* 4G/4G (18%). The *ACE* I/D polymorphism was the only significantly different factor among the cases and controls. The dominant genetic model D/D+I/D *vs* I/I showed an OR (95%CI) of 2.89 (1.22-6.89) and P = 0.019 in Mexican-Mestizo women. The results of this study support an association between the *ACE* I/D polymorphism and IRPL risk in a Mexican population.

Key words: *ACE I/D* polymorphism; Gene-disease association; Hypofibrinolysis; Recurrent pregnancy loss; Thrombophilia