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Association between the thrombophilic polymorphisms *MTHFR* C677T, Factor V Leiden, and *prothrombin* G20210A and recurrent miscarriage in Brazilian women

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ABSTRACT. Some cases of recurrent first trimester miscarriage have a thrombotic etiology. The aim of this study was to investigate the prevalence of the most common thrombophilic mutations - factor

V (*FV*) Leiden G1691A (*FVL*), prothrombin (*FII*) G20210A, and methylenetetrahydrofolate reductase (*MTHFR*) C677T - in women with recurrent miscarriages. In this case-control study, we included 137 women with two or more consecutive first-trimester miscarriages (£12 weeks of gestation) and 100 healthy women with no history of pregnancy loss, and with at least one living child. DNA was extracted from the patient samples, and the relevant genes (*FVL*, *FII*, and *MTHFR*) were amplified by PCR, followed by restriction fragment length polymorphism, to assess the polymorphisms in these genes. The allelic frequencies of polymorphisms were not significantly different between the case and control groups. Polymorphisms in the *MTHFR*, *FVL*, and *FII* genes were not associated with recurrent miscarriage during the first trimester of pregnancy in Brazilian women (P = 0.479; P = 0.491 and P = 0.107, respectively). However, the etiologic identification of genetic factors is important for genetic counseling.

Key words: Recurrent miscarriage; First trimester miscarriage; Factor V Leiden; Methylenetetrahydrofolate reductase C677T; *FII* G20210A