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TP53 gene polymorphisms at codons 11, 72, and 248 and association with endometriosis in a Brazilian population

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ABSTRACT. We evaluated the association between *TP53* gene polymorphisms and endometriosis in Brazilian women. Genomic DNA was extracted from swabs of buccal cells collected from hospital patients. *TP53* gene polymorphisms were investigated at three codons: *TP53**11 Glu/Gln or Lys (GAG->CAG or AAG), *TP53**72 Arg/Pro (CCG->CCC), and *TP53**248 Arg/Thr (CGG->TCG) using the polymerase chain reaction-restriction fragment length polymorphism method. *TP53**11 presented the following genotypic distribution: the control group was 98.28% homozygous wild-type (Glu) and 1.72% homozygous variant (Gln/Lys), and the heterozygous genotype was not identified. The genotypic distribution in the endometriosis group was 96% homozygous wild-type (Glu) and 4% heterozygous (Glu-Gln/Lys); the homozygous variant genotype was not identified (P = 0.02). *TP53**72 showed the following genotypic distribution: the control group was 29.75% homozygous wild-type (Arg), 47.11% heterozygous (Arg-Pro), and 23.14% homozygous variant (Pro). The genotypic

distribution in the endometriosis group was 16.15% homozygous wild-type (Arg), 51.54% heterozygous (Arg-Pro), and 32.31% homozygous variant (Pro) (odds ratio = 2.26; 95% confidence interval = 1.19-4.03; P = 0.02). Only one patient had the homozygous *TP53**248 genotype (Arg-Trp/Gln); all other patients were homozygous wild-type in both the control and endometriosis groups (P = 0.51; NS). We found that *TP53**72 polymorphism may be associated with susceptibility to endometriosis; the presence of at least 1 polymorphic allele increased the chance of disease development by 2.26-fold. Hence, this genetic variant is a potential candidate marker for endometriosis.

Key words: Cell cycle; Endometriosis; Gene polymorphism; p53; *TP53*

