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

C.M. Camargo-Kosugi, P. D’Amora, J.P.F.O. Kleine, C.V. Carvalho, H. Sato, E. Schor and I.D.C.G. Silva

Laboratório de Ginecologia Molecular e Proteômica, Departamento de Ginecologia, Escola Paulista de Medicina, Universidade Federal de São Paulo, São Paulo, SP, Brasil

Corresponding author: C.M. Camargo-Kosugi
E-mail: cintiakosugi@gmail.com

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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