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Association between male infertility and androgen receptor mutations in Brazilian patients

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ABSTRACT. The androgen receptor is encoded by a single-copy gene located in the long arm of the X chromosome (Xq11-12); it consists of eight exons and encodes an intracellular transcription factor that belongs to the steroid/nuclear receptor superfamily. Disturbances in the function of the androgen receptor can lead to several forms of male pseudohermaphroditism, such as androgen insensitivity syndrome, which can lead to infertility. Infertility affects around 20% of couples, and in half of the cases it is a male problem. Seventy male patients with idiopathic infertility were selected; data were obtained on age, drinking and smoking habits, occupation, and family history. The mean age of the patients was 37 years old (standard deviation = 12.3); 44% were azoospermic, 33% were oligozoospermic and 24% did not have alterations in the spermogram. Our objective was to evaluate a possible association between male infertility and mutations in the androgen receptor gene based on the presence or absence of exons 1 and 4 of this gene. These two exons were tested by PCR, and their products were separated on 1.5% agarose gels. We found that azoospermic patients had higher mutation rates on exons 1 and 4 of the androgen receptor gene, when compared to other alterations that

also lead to infertility, such as oligozoospermia and teratozoospermia. So, we conclude that patients who do not produce sperm have a higher number of mutations in the androgen receptor gene when compared to those who only have impaired sperm production. Based on molecular analysis, we found that there was no correlation between alterations in the spermogram and mutations on exons 1 and 4 of the androgen receptor gene and no association between alterations in the spermogram and alcohol drinking or smoking.

Key words: Male infertility; Androgen receptor; Exon 4; Exon 1; Chromosome X

