Absence of the exon 1 coding sequence of the androgen receptor gene associated with teratozoospermia in a Brazilian population

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ABSTRACT. The androgen receptor (AR) is a protein encoded by the AR gene, which when mutated may affect spermatogenesis, the process in which spermatozoa are produced; thus, AR mutations could lead to male infertility. We examined exon 1 of the AR gene in men with idiopathic infertility. Blood or semen samples from 111 infertile, oligozoospermic (N = 31), asthenozoospermic (N = 23), teratozoospermic (N = 33), and azoospermic (N = 24) men were analyzed. The extracted DNA was amplified for the exon 1 region of the AR gene. There was a significant correlation between the absence of exon 1 in the AR gene and spermatogenesis defects (P = 0.015). This association was significant in teratozoospermic men (51.5% of the sample). We found that lack of amplification of exon...
1 of the AR gene by polymerase chain reaction is associated with morphological defects in the spermogram.

**Key words:** Androgen receptor; AR gene; Male infertility; Spermatogenesis; Teratozoospermia