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FSB RECEPTOR GENE POLYMORPHISMS IN FERTILE AND INFERTILE MEN FROM EASTERN SICILY

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Background: Follicle-stimulating honnone (FSH) regulates spennatogenesis by a specific receptor (FSHR). Two single nucleotide polymorphisms (SNP) in exon IO of FSHR gene influence FSHR sensirivity in women: Thr307Ala (T307Ala) and Asn680Ser (N680S). In contrast, no effects ofthese SNPs on maJe serum FSH and spermatogenesis have been proven. Objecrives: The aim of this study was to eva.Iuate the frequency distriburion of the FSHR polymorphisms in infertile men from Eastem Sicily and their role on serum FSH levels. Subjects and methods: The SNPs were analyzed in 48 men with oligoasthenozoospennia (OAT) and in 33 normozoospermic controls by direct automated DNA sequencing of every PCR product containing the specific SNP. Results: An a.lmost complete linkage disequilibriurn was detected between positions 307 and 680, except for one patient. Their genotype frequencies were not significantly ditferent between OAT men [22.9% (Thrffhr-Asn!Asn), 56.3% {Thr/Ala-Asn!Ser) and 20.8% {Ala!Ala-Ser/Ser)] and fertile men [36.4%, 42.4% and 2.1.2%, respectively]. The FSHR genotypes did not result in different serum FSH and testosterone concentrations both in nonnozoospennic men and in men with OAT. The frequency distribution ofT307A and N680S genotypes in Sicilian men (28.4% for TNtrN, 50.6% TN/AS and 21% for AS/AS) was not statistically ditferent from that reported in men from Tuscany (for T307A: 29.7% (TT), 46.5% (TA) and 23.8% (AA) and for N680S: 30.7% {NN), 43.6% (NS), 25.7% (SS) (lnternarional HapMap Project). Conclusions: The FSHR gene Thr307Ala and Asn680Ser polymorphi sms were not differently distribuited in Sicilian men with oligozoospermia and normozoospennia and did not correlate with serum FSH concentrations. The heterozygous genotype TN/AS was the most represented.