Objective
The study of single nucleotide polymorphisms (SNPs) in candidate genes that regulate reproductive function may help to clarify the mechanisms responsible for gonadal function and fertility in humans. We aimed to evaluate SNPs in FSHR, AMH, AMHR2, ESR1, ESR2, CYP17A1, CYP19A1, COMT, MTHFR, BMP15, GDF9, FOXL2, LHX8 and NANOS3 genes as predictors of response to controlled ovarian hyperstimulation (COH) and in vitro fertilization (IVF) outcomes.

Design
Cross-sectional study.

Material and Methods
188 normovulatory women (32.5±3.4 years old) with infertility mainly caused by male, tuboperitoneal or idiopathic factors underwent IVF treatment were studied. FSH, estradiol and AMH levels, as well as antral follicle counting (AFC) were evaluated on 2nd or 3rd day of the menstrual cycle. Genotyping were performed by real-time PCR using TaqMan assay.

Results
GG genotype of AMHR2 rs11170555/G>A was associated lower AMH levels, while CC genotype of MTHFR rs1801131/A>C to higher AMH levels and AFC, and TT genotype of MTHFR rs1801133/C>T to lower AFC. Considering the response to COH, TT genotype of AMH rs10407022/G>T and GA of ESR2/rs4486938/G>A were more frequently found in satisfactory ovarian response, while GA genotype of AMHR2 rs3741664/G>A and CC genotype of MTHFR rs1801131/T>C in excessive response. The combination AMHR2/rs3741664 and MTHFR/rs1801131 or AMHR2/rs3741664 and ESR2/rs4486938 increases the chance of excessive response. AA genotype of FSHR/rs6165 was associated to higher numbers of oocytes retrieved and MII, meanwhile AA genotype of AMHR2/rs11170555; CC of AMHR2/rs2071558; GG of AMHR2/rs3741664 and TT genotype of AMHR2/rs2002555 were associated to lower numbers of oocytes retrieved and MII.

Conclusions
Genetic variation in AMH rs10407022 and ESR2 rs4986938 polymorphisms are associated to good response, meanwhile, AMHR2 rs3741664 and MTHFR rs1801131 polymorphisms are associated to excessive response to controlled ovarian hyperstimulation. Besides, these SNPs also influenced number of retrieved oocytes and MII in Brazilian infertile women.

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