

INVESTIGATION ON -211 G>T POLYMORPHISM ON GENE FSH β

AMPLI SET FSH β

Cat. N. 1.509RT

The *Follicle Stimulating Hormone*, or FSH, is a glycoprotein hormone generated and secreted by the adenohypophysis and contributes, in both sexes, to the regulation of the developing, puberal maturation and reproductive processes.

FSH is an heterodimer consisting in two sub-units, the α (shared with other glycoprotein hormone such as LH, hCG and TSH) and the sub-unit β that gives specificity to the activity. The sub-unit β is codified in the gene FSH β (4,2 kb), position 11p13 and having 3 codifying exons. The resulting protein sub-unit β consists in 123 amino acids. The inactivating mutations of β FSH are 5 and they can be found in the exon 3, causing a truncated protein or the loss of cysteine residual. The polymorphism -211 G>T (rs 10835638) affects the FSH level in the serum. This polymorphism causes the substitution of a G with a T and it is collocated in in the gene promotor at -211bp upstream from the transcriptional mRNA beginning site. It has been noticed a significant statistic association between FSH levels in the serum and the genotype of FSH β : heterozygous subjects (GT) or homozygous (TT) have serum level of FSH much lower than WT subjects WT (GG). Many studies, including Tüttelmann in 2012, have reveled a significant correlation between the subjects carrying allele T with serum FSH level (24% lower in TT than GG) and the rate FSH/LH, testicular volume, and concentration and spermatic count (36% lower and 34% lower TT than GG).

The assay allows the identification of the -211 G>T substitution in the FSH β gene promotor with molecular biology technique based on Real-time PCR.

Principle: A) extraction of genomic DNA B) amplification and detection Real-Time PCR. C) revelation with Real-Time PCR

Applicability: On extracted and purified genomic DNA from whole blood

Number of reactions: 50

REAGENTS AND STORAGE

AMPLIFICAZIONE	
PCR mix 5X	-20°C
Primer probe mix FSH β -211 G>T 20X	-20°C in the dark
H ₂ O RNase/DNase-FREE	-20°C
Heterozygous control GT	+4°C
Control GG	+4°C
Control TT	+4°C

Stability: over 12 months if correctly stored.

References:

Ann of Hum Gen (2007)71, 18-28
Endocrinology (2013) 154,3016-3021
Mol Hum Reprod (2002) 8, 893-899
JCEM (2012) 97, 3639-3647
JCEM (2010) 95, 100-108

ANALYSIS OF RESULTS

