

# FSHR (A2039G) N680S POLYMORPHISM

## ORDERING INFORMATIONS

REF: GEN-021-25 RDM Code: 1730074/R  
 Tests: 25 Reactions: 31  
 CND Code: W0106010499  
 Manufacturer: BioMol Laboratories s.r.l.

## CONTENTS OF THE KIT

The kit consists of: reagents for Real-Time PCR amplification  
 \*reagents for the extraction of genomic DNA are not supplied in the kit

For in vitro diagnostic use



## PRODUCT CHARACTERISTICS

Detection of N680S polymorphism of the FSHR gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx.

## SCIENTIFIC BACKGROUND

The physiological action of the FSH hormone depends on the activation of its receptor (FSHR). The FSH receptor is encoded by the FSHR gene located on chromosome 2p21-p16. Inactivating mutations of the FSHR gene have been described, but also multiple gene polymorphisms (about 900). The most common are the rs6165 and rs6166 polymorphisms, which correspond to the FSHR substitutions Thr307Ala and Asn680Ser respectively.

These polymorphisms have been extensively studied and it has been shown that the FSHR genotype related to these SNPs is predictive of ovarian responsiveness to FSH treatment. The analysis of the genotype of the FSH receptor therefore allows to individually modulate the administration of FSH and therefore to increase the efficacy and safety of the therapy.

Furthermore, many scientific works have recently been published on the correlation between FSH receptor polymorphisms (FSHR) and the risks of non-physiological spermatogenesis, correlating them with a functional deficit in the spermatogenesis process and therefore with a possible contributing cause in the phenomena of oligospermia or azoospermia.

## CLINICAL SIGNIFICANCE

Follicle Stimulating Hormone (FSH) performs its ovarian function through important effects on granulosa cell proliferation, oocyte maturation and estrogen synthesis. Multiple studies have shown that a decrease in FSH concentration followed by a high concentration of estrogen plays an important role in the selection of the dominant follicle. In humans, on the other hand, FSH is important for the regulation of the metabolic functions of Sertoli cells, an essential stage for the maintenance of normal spermatogenesis from a qualitative and quantitative point of view.

§ The susceptibility of FSHB -211G > T and FSHR G-29A, 919A > G, 2039A > G polymorphisms to men infertility: an association study and meta-analysis. *BMC Med Genet.* 2017 Aug 1; 18 (1):81.

§ FSH receptor gene p. Thr307Ala and p. Asn680Ser polymorphisms are associated with the risk of polycystic ovary syndrome. *J Assist Reprod Genet.* 2017 Aug; 34 (8):1087-1093. Epub 2017 May 25.

§ Follicle-Stimulating Hormone Receptor (FSHR): A Promising Tool in Oncology? *Mol Diagn Ther.* 2016 Dec; 20 (6):523-530. Review

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DESCRIPTION	LABEL	VOLUME	STORAGE
		<b>GEN-021-25</b>	
Mix oligonucleotides and probes	Mix N680S FSHR 10X	1 x 85 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 425 µl	-20°C
Deionized H <sub>2</sub> O	Deionized H <sub>2</sub> O	2 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control + 1	1 x 22µl	-20°C
Genomic DNA or recombinant DNA	Control + 2	1 x 22µl	-20°C
Genomic DNA or recombinant DNA	Control + 3	1 x 22µl	-20°C

## TECHNICAL CHARACTERISTICS

### COD. GEN-021-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
POSITIVE CONTROL	Recombinant DNA for at least 3 analytical sessions
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx e Agilent AriaDx
TECHNOLOGY	Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels
RUNNING TIME	85 min
THERMAL CYCLING PROFILE	1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec)
ANALYTICAL SPECIFICITY	Absence of non-specific pairings of oligonucleotides and probes; absence of cross-reactivity
ANALYTICAL SENSITIVITY : LIMIT OF DETECTION (LOD)	≥ 0,016 ng of DNA
ANALYTICAL SENSITIVITY : LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%