

POLYMORPHISM N312S (G935A) LHCGR

ORDERING INFORMATIONS

REF: GEN-026-25
 Tests: 25 Reactions: 31
 REF: GEN-026-50
 Tests: 50 Reactions: 62
 Manufacturer: BioMol Laboratories s.r.l.

CONTENTS OF THE KIT

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



PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Determination of the N312S (G935A) polymorphism of the LHCGR gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

Gonadotropins are hormones capable of regulating the activity of the gonads or, more simply, the functions of the male and female reproductive organs. The best known gonadotropins are two, called, respectively, LH (luteinizing hormone) and FSH (follicle stimulating hormone) and are secreted by the adenohypophysis. The latter is controlled by the hypothalamus, through a mechanism regulated by the hormone GnRH (gonadotropin-releasing hormone), which promotes the release of gonadotropins.

Once secreted, LH and FSH are transported by the blood to the target cells, on which specific membrane receptors are present. By interacting with them, they trigger a series of biochemical and metabolic events that differ depending on the organ involved.

The LHCGR gene, receptor for the LH hormone, is made up of 11 exons and is located on chromosome 2. One of the most studied polymorphic sites of the LHCGR gene is the c.935 G>A (AGT>AAT pS312N) variant (rs2293275) in 'exon 10. In Caucasian populations, approximately 18% are homozygous for N312, 49% are heterozygous (N312S), and 33% are homozygous for S312.

§ Effect Modification of LHCGR Gene Variant (rs2293275) on Clinico-Biochemical Profile, and Levels of Luteinizing Hormone in Polycystic Ovary Syndrome Patients. *Biochemical Genetics* (2023) 61:1418–1432 <https://doi.org/10.1007/s10528-022-10327-z>

§ Genetic Variants of Gonadotropins and Their Receptors Could Influence Controlled Ovarian Stimulation: IVF Data from a Prospective Multicenter Study. *Genes* 2023, 14, 1269.

§ A Comprehensive Overview of Common Polymorphic Variants in Genes Related to Polycystic Ovary Syndrome. *Reprod. Sci.* (2021) 28:2399–2412

§ Gonadotropin receptor variants are linked to cumulative live birth rate after in vitro fertilization. *Journal of Assisted Reproduction and Genetics* (2019) 36:29–38

§ LHCG receptor polymorphisms in PCOS patients. *BIOTECHNOLOGY & BIOTECHNOLOGICAL EQUIPMENT*, 2018 VOL. 32, NO. 2, 427–432

§ Combined assessment of polymorphisms in the LHCGR and FSHR genes predict chance of pregnancy after in vitro fertilization. *Human Reproduction*, Vol.31, No.3 pp. 672–683, 2016 Advanced Access publication on January 14.

§ Association of Luteinizing Hormone Chorionic Gonadotropin Receptor Gene Polymorphism (rs2293275) with Polycystic Ovarian Syndrome: GENETIC TESTING AND MOLECULAR BIOMARKERS Volume 19, Number 3, 2015

CLINICAL SIGNIFICANCE

The LHCGR N312S variant has not been explored to the same extent as the FSHR N680S variant in the context of controlled ovarian stimulation (COH) before IVF. However, it has recently been shown that subjects homozygous for S (homozygous GG) in both polymorphisms (FSHR and LHCGR) are four times more likely to achieve a pregnancy after the first IVF cycle, compared to women with other genotypes.

Furthermore, the rs2293275 polymorphism of the LHCGR gene has been associated with polycystic ovary syndrome (PCOS), which is a very common heterogeneous disease characterized by multiple clinical features including menstrual disorders, infertility, hirsutism, acne, insulin resistance and greater cardiovascular risk. Compared to GG homozygous subjects, a greater risk of developing PCOS was observed in subjects carrying the GA or AA genotypes.

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-026-25	GEN-026-50	
Mix oligonucleotides and probes	Mix N312S LHCGR 10X	1 x 85 µl	1 x 170 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 425 µl	1 x 850 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	2 x 1 ml	2 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control + 1	1 x 22 µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control + 2	1 x 22 µl	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control + 3	1 x 22 µl	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-026-25 / COD. GEN-026-50

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 and 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 pairing of oligonucleotides and probes; absence of cross-reactivity
ANALYTICAL SENSITIVITY : LIMIT OF DETECTION (LOD)	≥ 0,016 of genomic DNA
ANALYTICAL SENSITIVITY: LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%