ESR2 (A1730G) *39 AG POLYMORPHISM

For in vitro diagnostic use

CE IVD

ORDERING INFORMATIONS

REF: GEN-022-25 RDM Code: 1730075/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*

PRODUCT CHARACTERISTICS

Detection of A1730G (*39 A/G) polymorphism of the ESR2 gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

For the ESR1 and ESR2 genes there are multiple SNPs whose genotypic combinations explain the variability of the receptors in terms of quality and quantity. For the ESR1 gene (6q25) the most studied polymorphism is the T/C-397 polymorphism (rs2234693) localized in intron 1 of the gene. Such polymorphism is also called PvuII polymorphism, classified as Pp, depending on the presence or absence of the restriction site. The T nucleotide is also termed the p allele, while the C nucleotide is termed the P allele. The PP genotype (CC) is associated with receptor dysfunction with impaired response to estrogen. For the ESR2 gene, the most studied polymorphism is located in the 3'UTR region of the gene, at the level of nucleotide 1730 (1730 A→G) (rs4986938), and is recognized by the restriction enzyme AluI. This polymorphism is also known as *39 A→G. The *39GG genotype is associated with a reduced response to estrogen. The presence of these polymorphic variants represents a susceptibility factor for multiple conditions such as the risk of developing cancer (breast, colorectal, prostate cancer), neurodegenerative diseases (e.g. Parkinson's, Alzheimer's) and the couple's fertility status.

§ Differential association of ESR1 and ESR2 gene variants with the risk of breast cancer and associated features: A case-control study. Gene. 2018 Apr 20; 651:194-199. Epub 2018 Feb 4.

§ Polymorphisms in the estrogen receptor alpha gene (ESR1), daily cycling estrogen and mammographic density phenotypes.BMC Cancer. 2016 Oct 7; 16(1):776.

§ A Study on the Role of Estrogen Receptor Gene Polymorphisms in Female Infertility. Genet Test Mol Biomarkers. 2016 Nov; 20 (11):692-695. Epub 2016 Aug 30.

CLINICAL SIGNIFICANCE

Estrogen receptors (ERs) are members of the large superfamily of ligand-activated nuclear receptors. To date, two receptor isoforms have been identified: ER-α (ESR1 gene) and ER-β (ESR2 gene). Both receptors belong to the nuclear receptor superfamily, but are synthesized by different genes and have unique structures and functions. The two isoforms consist of six domains and show high sequence homology (96%) in the DNA binding region, while they have distinct structures in the site of interaction with ligands (53% homology). The ESR1 gene is located on chromosome 6 and encodes the ER-α protein, abundantly expressed in the liver, adipose tissue, breast and cardiovascular system. Activated ER-α receptor has been shown to regulate the hepatic expression of many genes involved in lipoprotein metabolism, resulting in increased serum HDL cholesterol (HDL) and triglyceride concentrations while decreasing serum low-density lipoprotein and cholesterol lipoprotein (LDL). The ESR2 gene encodes the ER-β protein and is located on chromosome 14q23.1. ER-β is expressed in many tissues including the uterus, tissue monocytes and macrophages, colonic and lung epithelial cells, and in the prostatic epithelium and in the malignant counterparts of these tissues. Furthermore, ER-β is expressed throughout the brain at different concentrations in relation to neuronal areas.

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ISO 9001:2015
ISO 13485:2016

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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-022-25	
Mix oligonucleotides and probes	Mix *39 A/G ESR2 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_20	Deionized H_20	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-022-25

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%

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