

ESR1 -397 T>C POLYMORPHISM (ESTROGEN RECEPTOR)

ORDERING INFORMATIONS

REF: GEN-023-25 RDM Code: 1734263/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

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Detection of -397 T/C polymorphism of the ESR1 (rs2234693) gene by Real-Time PCR technique. Kit optimized for Real-Time PCR instruments Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

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.

CLINICAL SIGNIFICANCE

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 polymorphism-397 T/C (rs2234693) located in intron 1 of the gene. Such a polymorphism is also called a PvuII polymorphism, classified as a Pp, depending on the presence or absence of the restriction site. Nucleotide T is also referred to as the p allele, while nucleotide C is referred to as the P allele. The PP genotype is associated with receptor dysfunction with reduced response to endogenous estrogens. Furthermore, the presence of this polymorphic variant represents a susceptibility factor for multiple conditions such as the risk of developing tumor pathologies (breast cancer, colorectal cancer, prostate cancer), neurodegenerative diseases (e.g. Parkinson's, Alzheimer's) and fertility status of the couple. Positive interactions have been observed between ESR2 rs4986938, ESR1 rs2234693 and triple negative breast cancer (TNBC).

- § Diagnostics (Basel). 2024 Aug 28;14(17):1889. Association of Polymorphisms in FSHR, ESR1, and BMP15 with Primary Ovarian Insufficiency and Meta-Analysis
- § Meta-Analysis Cancer Genomics Proteomics. 2024 Sep-Oct;21(5):421-438. Pharmacogenetics of Toxicities Related to Endocrine Treatment in Breast Cancer: A Systematic Review and Meta-analysis
- § In Vivo. 2024 Sep-Oct;38(5):2134-2143. Analysis of Single Nucleotide Polymorphisms (SNPs) rs2234693 and rs9340799 of the ESR1 Gene and the Risk of Breast Cancer
- § Urol J. 2024 Jun 12. Association of Polymorphisms in Estrogen Receptors with non-obstructive Azoospermia and Severe Secretory Oligozoospermia: Meta-Analysis
- § Gene. 2023 Jan 30;851:146969. Unique ESR1 and ESR2 estrogen receptor gene variants associated with altered risk of triple-negative breast cancer: A case-control study
- § ESR1 PvuII polymorphism: from risk factor to prognostic and predictive factor of the success of primary systemic therapy in advanced breast cancer. BMC Cancer volume 21, Article number: 1348 (2021)
- § BMC Cardiovasc Disord. 2021 Jun 4;21(1):275. The association between estrogen receptor 2 gene polymorphism and complexity of coronary artery disease: an analysis in elective percutaneous coronary intervention patients
- § Medicine (Baltimore). 2021 Feb 19;100(7):e24398. The role of estrogen receptor-beta gene +1730C/A polymorphisms in recurrent pregnancy loss: A protocol for systematic review and meta-analysis
- § 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.

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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-023-25	
Mix oligonucleotides and probes	Mix -397 T/C ESR1 10X	1 x 77,5 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 387,5 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control 1	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 3	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-023-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissues, cells
CONTROLS	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 pairings of oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 0,016 ng of genomic DNA
LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%