

GENETIC VARIANT ARG399GLN OF THE XRCC1 GENE

ORDERING INFORMATION

REF: FGC-011-25
RDM Code: 2259495/R
CND Code: W0106010499
Tests: 25 Reactions: 31
Manufacturer: BioMol Laboratories s.r.l.

CONTENTS OF THE KIT

The kit consists of reagents for Real-Time PCR amplification
*the 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**. Determination of the ARG399GLN polymorphism of the XRCC1 gene (G>A; ARG399GLN; rs25487) by amplification with oligonucleotides and specific probes (allele-specific genotyping) and subsequent detection with qPCR-Real-time. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

SCIENTIFIC BACKGROUND

Radiation therapy is a potentially curative and important treatment option in the early stages of localized cancer. Radiation therapy and cytotoxic treatment destroy cancer cells by inducing DNA damage. Therefore, the outcome of these treatments depends on the effectiveness of the DNA repair systems. The XRCC1 (X-Ray repair cross complementing group 1) protein is essentially involved in both single-strand break repair and base excision repair. The single nucleotide polymorphism (SNPs) of the XRCC1(rs25487) gene identifies the G>A substitution that causes the variation of codon 399 of the amino acid arginine (Arg) to the amino acid glutamine (Gln).

§ Mol Cells. 2025 Jan 17;100186. doi: 10.1016/j.mocell.2025.100186. Online ahead of print. Cancer prognosis using base excision repair genes

§ Biomol Biomed. 2024 Dec 13. doi: 10.17305/bb.2024.11314. Online ahead of print. The association of rs25487 of the XRCC1 gene and rs13181 of the ERCC2 gene polymorphisms with the ovarian cancer risk

§ Front Pharmacol. 2024 Aug 21;15:1445328. doi: 10.3389/fphar.2024.1445328. eCollection 2024. Genetic polymorphisms and platinum-induced hematological toxicity: a systematic review

§ BMC Cancer. 2024 Jan 15;24(1):78. Novel model integrating computed tomography-based image markers with genetic markers for discriminating radiation pneumonitis in patients with unresectable stage III non-small cell lung cancer receiving radiotherapy: a retrospective multi-center radiogenomics study

§ Reprod Sci. 2023 Apr;30(4):1118-1132. Elucidation of Increased Cervical Cancer Risk Due to Polymorphisms in XRCC1 (R399Q and R194W), ERCC5 (D1104H), and NQO1 (P187S)

§ Nucleosides Nucleotides Nucleic Acids. 2022;41(5-6):530-554. Association of genetic polymorphisms in DNA repair genes ERCC2 Asp312Asn (rs1799793), ERCC2 Lys 751 Gln (rs13181), XRCC1 Arg399 Gln (rs25487) and XRCC3 Thr 241Met (rs861539) with the susceptibility of lung cancer in Saudi population

§ Front Oncol. 2021 May 19;11:654784. Significant Association Between XRCC1 Expression and Its rs25487 Polymorphism and Radiotherapy-Related Cancer Prognosis

§ J Cell Biochem. 2017 Dec;118(12):4782-4791. Evaluation of Prediction of Polymorphisms of DNA Repair Genes on the Efficacy of Platinum-Based Chemotherapy in Patients With Non-Small Cell Lung Cancer: A Network Meta-Analysis

§ XRCC1 rs25487 Polymorphism Predicts the Survival of Patients After Postoperative Radiotherapy and Adjuvant Chemotherapy for Breast Cancer ANTICANCER RESEARCH 34: 3031-3038 (2014)

§ Genetic polymorphisms in XRCC1 associated with radiation therapy in prostate cancer Cancer Biology & Therapy 10(1), 13-18; July 1, 2010.

§ Functional characterization of polymorphisms in DNA repair genes using cytogenetic challenge assays. Environ Health Perspect 111: 1843-1850, 2003. ANTICANCER RESEARCH 34: 3031-3038 (2014) 3036

CLINICAL SIGNIFICANCE

Studies have been conducted on the functional effects of the amino acid substitution Arg399Gln, suggesting that the genotype of the AA variant is associated with a 3- to 4-fold reduced DNA repair capacity.

In addition, it has also been associated with an increase in chromosomal deletions, increasing the risk of cancer. Recent meta-analysis studies have shown that polymorphisms in the DNA damage repair genes XRCC1 (rs25487 and rs1799782), ERCC5 (rs17655) and the oxidative stress-related NQO1 gene (rs1800566) are significantly associated with an increased risk of developing cancer. Recently, an increased risk of developing ovarian cancer has been demonstrated in subjects carrying the GA and AA genotypes of the rs25487 polymorphism.

DNA repair genes increase susceptibility to lung cancer (LC) occurrence in the Saudi population through gene-gene interaction rather than through independent variants.

On the other hand, the data indicate that in terms of overall response ratio (ORR), ERCC1 (rs11615), XRCC1 (rs25487, rs1799782), and XPD (rs13181) polymorphisms are associated with the efficacy of platinum-based chemotherapy in non-small cell lung cancer (NSCLC).

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DESCRIPTION	LABEL	VOLUME	STORAGE
		FGC-011-25	
Mix oligonucleotides and probes	Mix 10X Arg399Gln XRCC1	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. FGC-011-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
CONTROLS	Recombinant DNA for at least 3 analytical sessions
TECHNOLOGY	Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx
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%