GENETIC VARIANT ARG399GLN OF THE XRCC1 GENE

For in vitro diagnostic use

CE IVD

ORDERING INFORMATIONS

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.*

PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-TESTS IN PHARMACOGENETICS.** Determination of the G399A G>A polymorphism of the XRCC1 gene (ARG399GLN) 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

Radiotherapy is a potentially curative and important therapeutic option in the early stages of localized carcinoma. Radiotherapy and cytotoxic treatment destroy cancer cells by inducing DNA damage. Therefore, the outcome of these treatments depends on the effectiveness of 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 G399A G>A mutation and involves the substitution in codon 399 of the amino acid arginine (Arg) to the amino acid glutamine (Gln).

§ 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 AA variant genotype is associated with a 3- to 4-fold reduced DNA repair capacity.
Furthermore, it has also been associated with an increase in chromosomal deletions, increasing cancer risk.
Recent meta-analysis study has demonstrated that polymorphisms in DNA damage repair genes XRCC1 (rs25487 and rs1799782), ERCC5 (rs17655), and oxidative stress-related gene NQO1 (rs1800566) are signifcantly associated with increased cervical cancer risk.
DNA repair genes increase susceptibility of lung cancer (LC) onset in the Saudi population via gene-gene interaction rather than independent variants.
On the other hand, 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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EN ISO 9001 CERTIFICATE
EN ISO 13485 CERTIFICATE

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DESCRIPTION	LABEL	VOLUME	STORAGE
		FGC-011-25	
Mix oligonucleotides and probes	Mix 10X Arg399Gln XRCC1	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 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
POSITIVE CONTROL	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 e 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
ANALYTICAL SENSITIVITY: LIMIT OF DETECTION (LOD)	≥ 0,016 ng of genomic DNA
ANALYTICAL SENSITIVITY: LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%

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