

CYP2C19 *2, *3 and *17 GENETIC VARIANTS

ORDERING INFORMATIONS

REF: FGC-019-25
Tests: 25
Reactions: 31 x 3
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

Device belonging to the family of in vitro devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS- RESEARCH USE ONLY**. Detection of genetic variants *2, *3, and *17 of the CYP2C19 gene (rs4244285, rs4986893, and rs12248560) by amplification with specific oligonucleotides and probes (allele-specific genotyping) and subsequent detection by qPCR-Real-time. Kit optimized for Real-time PCR instruments Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

The CYP2C19 enzyme contributes to the metabolism of a range of clinically important drugs, including antidepressants, benzodiazepines, voriconazole, some proton pump inhibitors, and the antiplatelet agent, clopidogrel.

The CYP2C19 gene is highly polymorphic, with over 35 variant star (*) alleles cataloged by the PharmVar Consortium.

CYP2C19 rs4244285 (c.681G>A) is the defining polymorphism of the CYP2C19*2 allele and is a synonymous G>A transition in exon 5 that creates an aberrant splice site. This change alters the mRNA reading frame, which results in a truncated, non-functional protein.

CYP2C19 rs4986893 (G>A) is a SNP in the CYP2C19 gene, encoding the CYP2C19*3 variant. Allele A is poor metabolizer of several commonly prescribed drugs.

CYP2C19 rs12248560 (c.-806C>T) is the defining polymorphism of the CYP2C19*17 allele and is a C>T transition in the promoter that creates a consensus binding site for the GATA transcription factor family, resulting in increased CYP2C19 expression and activity.

CLINICAL SIGNIFICANCE

The CYP2C19*1 is considered the normal allele and is categorized with normal enzyme activity and the Normal Metabolizer phenotype.

The CYP2C19*17 allele is associated with increased enzyme activity and, depending on the number of alleles present, is associated with the "rapid" (RM) (one *17 allele) or "ultrarapid" (UM) (*17/*17 diplotype) metabolizer phenotype.

Non-functional alleles include CYP2C19*2 and *3. The CYP2C19 IMs have one copy of an allele that encodes a non-functional enzyme (for example, *1/*2), whereas PMs have 2 non-functional alleles (for example, *2/*2, *2/*3). Individuals with one normal function allele and one decreased function allele are classified as having a "likely IM" phenotype.

§ **Meta-Analysis** Int J Cardiol. 2024 Nov 1;414:132418. Effect of loss-of-function CYP2C19 variants on clinical outcomes in coronary artery disease patients treated with clopidogrel.

§ **Meta-Analysis** JAMA Netw Open 2024 Aug 1;7(8):e2425593. Pharmacogenetic Variants and Plasma Concentrations of Antiepileptic Drugs: A Systematic Review and Meta-Analysis.

§ Eur Heart J Cardiovasc Pharmacother. 2024 Oct 4;10(6):526-536. Comparative effects of different antiplatelet strategies in carriers of CYP2C19 loss-of-function alleles: a network meta-analysis

§ **Meta-Analysis** Pharmacogenomics J. 2021 Apr;21(2):190-199. Global distribution of CYP2C19 risk phenotypes affecting safety and effectiveness of medications.

§ Review Clin Pharmacol Ther. 2021 Feb;109(2):352-366. PharmVar GeneFocus: CYP2C19

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DESCRIPTION	LABEL	VOLUME	STORAGE
		FGC-019-25	
Mix oligonucleotides and probes	Mix 10X CYP2C19 *2	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 10X CYP2C19 *3	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 10X CYP2C19 *17	1 x 62 µl	-20°C
Mix buffer and Taq-polymerase enzyme	Mix Real-Time PCR 2X	1 x 930 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA CYP2C19*2	Control 1, Control 2	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA CYP2C19*3	Control 1, Control 2	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA CYP2C19*17	Control 1, Control 2	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. FGC-019-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissues, cells
POSITIVE 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, Thermofisher QuantStudio™ 5 Real-Time PCR System.
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)	≥ 5 ng of genomic DNA
LIMIT OF BLANK (LOB)	>40 Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9%/98%