



SUPEROXIDE DISMUTASE 2 GENETIC VARIANT

ORDERING INFORMATIONS

REF: GEN-048-25
Tests: 25 Reactions: 31
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 the Ala16Val (C>T) polymorphism of the SUPEROXIDE DISMUTASE 2 GENE (rs4880; C47T) by Real-Time PCR technique. Kit optimized for Real-time PCR instruments Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx. Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

Superoxide dismutase 2 (SOD2), also known as manganese superoxide dismutase (MnSOD), is one of the major antioxidant defense systems against mitochondrial super oxide radicals.

Changes in the activity of the SOD2 antioxidant enzyme have been associated with altered progression and risk of different diseases.

Diabetes Mellitus (DM) is a chronic heterogeneous disorder and oxidative stress is a key participant in the development and progression of it and its complications. Anti-oxidant status can affect vulnerability to oxidative damage, onset and progression of diabetes and diabetes complications.

§ Genes Nutr. 2025 Jan 21;20(1):1. Impact of single nucleotide polymorphisms (SNPs) in antioxidant-enzyme genes on the concentrations of folate, homocysteine and glutathione in plasma from healthy subjects after folic acid supplementation - a randomized controlled crossover trial

§ Mol Biol Rep. 2024 Jul 13;51(1):805. Modifiable risk factors, oxidative stress markers, and SOD2 rs4880 SNP in coronary artery disease: an association study

§ Association of a single nucleotide polymorphism in SOD2 with susceptibility for the development of diabetic nephropathy in patients with type 2 diabetes: A Saudi population study. Endocrinol Diab Metab. 2023;6:e449. <https://doi.org/10.1002/edm2.449>

§ SOD2 gene Val16Ala polymorphism is associated with macroalbuminuria in Mexican Type 2 Diabetes patients: a comparative study and meta-analysis. Ascencio-Montiel et al. BMC Medical Genetics 2013, 14:110 <http://www.biomedcentral.com/1471-2350/14/110>

§ Role of Superoxide Dismutase 2 Gene Ala16Val Polymorphism and Total Antioxidant Capacity in Diabetes and its Complications. Avicenna Journal of Medical Biotechnology, Vol. 8, No. 2, April-June 2016

CLINICAL SIGNIFICANCE

The SOD2 gene is located on chromosome 6 in the q25 region. The genetic variant rs4880 results in a C>T substitution (GCT/GTT), within exon 2 of SOD2, resulting in an amino acid change from alanine to valine (Ala16Val), ultimately causing altered secondary structure and function of the enzyme. SOD2 enzyme activity is higher in CC subjects (Ala16Ala), while subjects with TT genotype (Val16Val) have lower enzyme activity, a result of less efficient targeting of the enzyme from the cytoplasm to the mitochondria.

Previous studies have reported that SOD2 (CT+TT) alleles are associated with 25% reduced catalytical activity of the enzyme, which are potentially disrupting the cellular redox homeostasis.

The TT genotype of this variant is associated with increased CHD (coronary heart disease) risk and lower plasma anti-oxidant defences in patients with diabetes. Recently it has been demonstrated that (CT+TT) alleles have lower concentrations of s-folate and higher concentration of p-tHcy than subjects with (CC) alleles.

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DESCRIPTION	LABEL	VOLUME	STORAGE
		GEN-048-25	
Mix oligonucleotides and probes	Mix SOD-2 rs4880 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 310 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-048-25

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, Thermofisher QuantStudio™ 5 Real-Time PCR System.
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 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%