

VITAMIN-D RECEPTOR GENETIC VARIANTS

ORDERING INFORMATION

REF: GEN-049-25
Tests: 25 Reactions: 31 x 4
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 medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS- RESEARCH USE ONLY**. Detection of the gene variants **BsmI (rs1544410, G/A)**, **FokI (rs2228570, C/T)**, **TaqI (rs731236, T/C)**, **Apal (rs7975232, A/C)** of the gene encoding the vitamin D receptor by Real-Time PCR technique. Kit optimized for Real-time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

Vitamin D is an important vitamin and steroid pro-hormone that plays a critical role in mineralization and bone metabolism. Regulates serum calcium (sCa) and serum phosphate (sP) concentrations, ensures adequate deposition of hydroxyapatite in the bone matrix and affects skeletal development, maintenance of skeletal architecture, on hormone secretion and immune function. This action of vitamin D is mediated by the vitamin D receptor (VDR) which binds specifically to 1,25 dihydroxyvitamin D3. Genetic discoveries have begun to explain the complex associations between the vitamin D pathway and bone health. In addition, alterations to the vitamin D receptor gene (VDR) play a key role in altering the absorption of vitamin D, potentially changing its function and increasing susceptibility to cardiovascular disorders.

§ Arch Dermatol Res. 2025 Feb 19;317(1):437. Role of VDR gene polymorphisms and environmental factors in the development of skin cancers: evidence by updated meta-analysis.

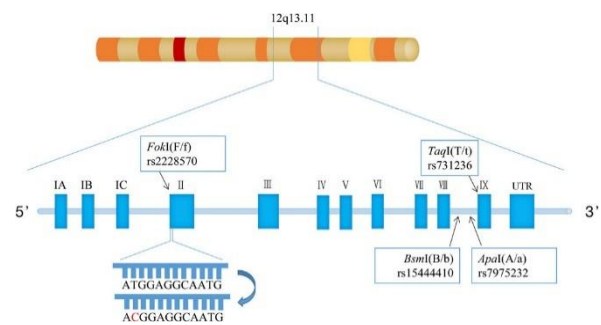
§ Acta Medica (Hradec Kralove). 2024;67(4):113-124. Updated Meta-Analysis of VDR FokI and TaqI Variants and Their Association with Melanoma Risk

§ Cureus. 2022 Dec 19;14(12): e32715. Vitamin D Receptor Gene Polymorphism Apal as a Predisposing Factor for Psoriasis and Its Relation With Serum Vitamin D Levels and Psoriasis Severity.

§ Meta-Analysis Nucleosides Nucleotides Nucleic Acids. 2023;42(1):17-37. Lower vitamin D levels and VDR variants are risk factors for breast cancer: an updated meta-analysis.

CLINICAL SIGNIFICANCE

The gene encoding the Vit receptor. D (VDR) is considered an important candidate for the genetic regulation of bone strength, bone homeostasis and metabolism. The gene, about 75 kb, is located on chromosome 12q13.11 and contains 12 exons and almost 300 different polymorphisms. The most studied polymorphisms of the VDR gene are four: Apal (rs7975232 A/C), BsmI (rs1544410 A/G), FokI (rs2228570 T/C), TaqI (rs731236, T/C).



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DESCRIPTION	LABEL	VOLUME GEN-049-25	STORAGE
Mix oligonucleotides and probes	Mix 1 (VDR BsmI G>A) 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 2 (VDR FokI C>T) 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 3 (VDR TaqI T>C) 10X	1 x 62 µl	-20°C
Mix oligonucleotides and probes	Mix 4 (VDR ApaI A>C) 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 5X	1 x 496 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control 1, Control 2, Control 3 BsmI G>A	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 1, Control 2, Control 3 FokI C>T	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 1, Control 2, Control 3 TaqI T>C	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 1, Control 2, Control 3 ApaI A>C	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-049-25

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
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)	>40Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9-98%