

PRUNE-1 and RPP-30 COPIES NUMBER DETECTION (CNV)

ORDERING INFORMATIONS

REF: *ONC-022-100*
Tests: *100 Reactions: 124*
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

Determination of the copy number (CNV) of the PRUNE-1 and RPP-30 genes by PCR (Polymerase Chain Reaction) technique and subsequent detection in Real-time PCR. Kit optimized for Biorad CFX96 Dx, Biorad Opus Dx and Agilent AriaDx Real-Time PCR instrumentation, ThermoFisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

Numerous studies have reported that genes associated with the malignant phenotype of multiple myeloma (MM) are located on amplicon 1q21, to which the PRUNE-1 gene maps. The PRUNE-1 gene is a member of the DHH (Asp-His-His) group of phosphodiesterases. PRUNE-1 has recently been shown to play a role in neuronal migration and proliferation, especially in microtubule polymerization. Due to its enzymatic activities and its interaction with various proteins, Prune-1 is able to modulate both intra- and extracellular signaling cascades [including the canonical Wnt and TGF- β pathways] that regulate cell proliferation, motility and epithelial-mesenchymal transition (EMT) processes. To date, Prune-1 has been found to be highly expressed and positively correlated with grading, EMT and metastatic status in several tumors, such as medulloblastoma, gastric carcinoma (GC), esophageal squamous cell carcinoma (ESCC), non-atrial lung cancer small cell cancer (NSCLC), thyroid cancer (TC), colorectal cancer (CRC), neuroblastoma and triple-negative metastatic breast cancer. Prune-1 overexpression occurs through amplification and/or gain of chromosome 1q.

- § PRUNE1 (located on chromosome 1q21.3) promotes multiple myeloma with 1q21 Gain by enhancing the links between purine and mitochondrion Xu et al Br J Haematol. 2023;00: 1-15
- § Perspectives on the Risk-Stratified Treatment of Multiple Myeloma Davies et al Blood Cancer Discov. 2022 Jul 6;3(4):273-284
- § Front Oncol. 2021 Oct 22;11:758146. Functional Genomics of PRUNE1 in Neurodevelopmental Disorders (NDDs) Tied to Medulloblastoma (MB) and Other Tumors
- § Gain/Amplification of Chromosome Arm 1q21 in Multiple Myeloma Hanamura. Cancers 2021, 13(2), 256
- § Revealing the impact of structural variants in multiple myeloma Rustad et al Blood Cancer Discov. 2020 Nov;1(3):258-273.
- § Cell of Origin and Genetic Alterations in the Pathogenesis of Multiple Myeloma. Barwick et al. Front Immunol. 2019 May 21;10:1121.
- § A copy number variation map of the human genome; Zarrei et al Nat Rev Genet. 2015 Mar;16(3):172-83.
- § Understanding h-prune biology in the fight against cancer, Marino et al. Clin Exp Metastasis. 2007;24(8):637-45.
- § Prune cAMP phosphodiesterase binds nm23-H1 and promotes cancer metastasis; D'Angelo et al Cancer Cell. 2004 Feb;5(2):137-49.

CLINICAL SIGNIFICANCE

Monitor disease progression on tumors solids and liquids (multiple myeloma, carcinoma breast, lung, bladder, hepatocellular carcinoma, skin tumors, brain tumors in adults and children, B-cell lymphomas) of the colon rectum, and gastric tract cancer, measuring the "copy number variation" of the gain region of 1q21.3 in PRUNE-1 gene, gene "drivers" of a pathway PI-3- activity driven metastatic signaling Kinase and mTOR and signaling inhibition of the tumor suppressor PTEN. This amplification gene is important in the prognosis phase of the tumor pathology. PRUNE-1 amplification is index of poor prognosis in breast cancer, e multiple myeloma.

The ONC-022 product allows the determination of related number of copies (CNV) of the PRUNE-1 gene to the number of copies of the Ribonuclease P protein gene subunit p30 (RPP-30), a housekeeping gene highly preserved and used as an "internal reference" using Real-Time PCR technique.

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DESCRIPTION	LABEL	VOLUME	STORAGE
		ONC-022-100	
Mix oligonucleotides and probes	Mix PRUNE-1/RPP-30 10X	4 x 77,5 µl	-20°C
Mix buffer and enzyme Taq polymerase	Mix PCR 5X	4 x 155 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Recombinant DNA	CTR1 Jukart genomic DNA	1 x 50 µl	-20°C
Recombinant DNA	CTR2 HeLa genomic DNA	1 x 50 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. ONC-022-100

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood or tissue.
POSITIVE CONTROL	Recombinant DNA for at least 3 analytical sessions
TECHNOLOGY	Real-time PCR; oligonucleotides and probes specific for the PRUNE-1 and RPP-30 genes; 2 FAM/HEX fluorescence channels
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System
RUNNING TIME	85 min
THERMAL CYCLING PROFILE	1 cycle at 95 °C (10 min); 50 cycles at 95 °C (15 sec) + 60 °C (1 min)
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	100%/98%