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WFS1 Rabbit pAb

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Catalog # AP54440

Product Information

Application	WB
Primary Accession	O76024
Reactivity	Human
Predicted	Rat, Dog, Mouse, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	100292
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human WFS1
Epitope Specificity	791-890/890
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Endoplasmic reticulum; endoplasmic reticulum membrane; multipass membrane protein
DISEASE	Defects in WFS1 are the cause of Wolfram syndrome type 1 (WFS1) [MIM:222300]. A rare autosomal recessive disorder characterized by juvenile diabetes mellitus, diabetes insipidus, optic atrophy, deafness and various neurological symptoms. Defects in WFS1 are the cause of deafness autosomal dominant type 6 (DFNA6) [MIM:600965]; also called non-syndromic sensorineural deafness autosomal dominant type 14 (DFNA14) or non-syndromic sensorineural deafness autosomal dominant type 38 (DFNA38). DFNA6 is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNA6 is a low-frequency hearing loss in which frequencies of 2000 Hz and below are predominantly affected. Many patients have tinnitus, but there are otherwise no associated features such as vertigo. Because high-frequency hearing is generally preserved, patients retain excellent understanding of speech, although presbycusis or noise exposure may cause high-frequency loss later in life. DFNA6 worsens over time without progressing to profound deafness. Defects in WFS1 are the cause of Wolfram-like syndrome autosomal dominant (WFSL) [MIM:614296]. A disease characterized by the clinical triad of congenital progressive hearing impairment, diabetes mellitus, and optic atrophy. The hearing impairment, which is usually diagnosed in the first decade of life, is relatively constant and alters mainly low- and middle-frequency ranges.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Wolfram syndrome protein (WFS1) is an 890 amino acid protein that contains a cytoplasmic N-terminal domain, followed by nine-transmembrane domains and a luminal C-terminal domain. WFS1 is predominantly localized to the endoplasmic reticulum (ER) (1) and its expression is induced in response to ER

stress, partially through transcriptional activation (2,3). Research studies have shown that mutations in the WFS1 gene lead to Wolfram syndrome, an autosomal recessive neurodegenerative disorder defined by young-onset, non-immune, insulin-dependent diabetes mellitus and progressive optic atrophy (4).

Additional Information

Gene ID	7466
Other Names	Wolframin, WFS1
Dilution	WB=1:500-2000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

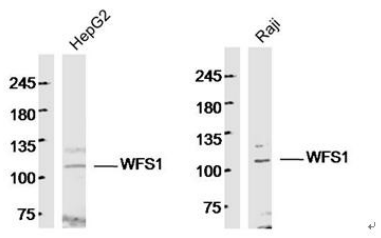
Protein Information

Name	WFS1
Function	Participates in the regulation of cellular Ca(2+) homeostasis, at least partly, by modulating the filling state of the endoplasmic reticulum Ca(2+) store (PubMed:16989814). Negatively regulates the ER stress response and positively regulates the stability of V-ATPase subunits ATP6V1A and ATP1B1 by preventing their degradation through an unknown proteasome-independent mechanism (PubMed:23035048).
Cellular Location	Endoplasmic reticulum membrane; Multi-pass membrane protein. Cytoplasmic vesicle, secretory vesicle. Note=Co-localizes with ATP6V1A in the secretory granules in neuroblastoma cell lines
Tissue Location	Highly expressed in heart followed by brain, placenta, lung and pancreas. Weakly expressed in liver, kidney and skeletal muscle. Also expressed in islet and beta-cell insulinoma cell line

Background

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Images



Sample:
HepG2 Cell (Human)Lysate at 40 ug
Raji Cell (Human)Lysate at 40 ug
Primary: Anti-WFS1 (AP54440) at 1/300 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 97 kD
Observed band size: 105 kD

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.