WFS1 Rabbit pAb

CatalogNo: YN3003

Key Features

Host Species
- Rabbit

Reactivity
- Human,Mouse

Applications
- WB,ELISA

MW
- 97kD (Observed)

Isotype
- IgG

Recommended Dilution Ratios

WB 1:500-2000
ELISA 1:5000-20000

Storage

Storage*	-15°C to -25°C/1 year(Do not lower than -25°C)
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Basic Information

Clonality	Polyclonal

Immunogen Information

Immunogen	Synthesized peptide derived from part region of human protein
Specificity	WFS1 Polyclonal Antibody detects endogenous levels of protein.

Target Information

Gene name	WFS1

Protein Name Wolframin

Organism	Gene ID	UniProt ID
Human	7466;	O76024;
Mouse		P56695;

Cellular Localization Endoplasmic reticulum membrane ; Multi-pass membrane protein . Cytoplasmic vesicle, secretory vesicle . Co-localizes with ATP6V1A in the secretory granules in neuroblastoma cell lines. .

Tissue specificity 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.

Function Disease:Defects in WFS1 are the cause of non-syndromic sensorineural 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.,Disease:Defects in WFS1 are the cause of Wolfram syndrome (WFS) [MIM:222300]; also known as diabetes insipidus and mellitus with optic atrophy and deafness syndrome (DIDMOAD). It is a rare autosomal recessive disorder characterized by juvenile diabetes mellitus, diabetes insipidus, optic atrophy, deafness and various neurological symptoms.,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.,polymorphism:Arg-456-His, Arg-611-His and Ile-720-Val polymorphisms are in tight linkage disequilibrium with one another and associated with type 1 diabetes in Japanese.,tissue specificity: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.,

Validation Data

Contact information

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Please scan the QR code to access additional product information:
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