WFS1 Monoclonal Antibody

Catalog No	BYmab-07847
Isotype	IgG
Reactivity	Human;Mouse
Applications	WB
Gene Name	WFS1
Protein Name	Wolframin
Immunogen	Synthesized peptide derived from part region of human protein
Specificity	WFS1 Monoclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	97kD
Cell Pathway	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
Background	This gene encodes a transmembrane protein, which is located primarily in the endoplasmic reticulum and ubiquitously expressed with highest levels in brain, pancreas, heart, and insulinoma beta-cell lines. Mutations in this gene are associated with Wolfram syndrome, also called DIDMOAD (Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy, and Deafness), an autosomal recessive disorder. The disease affects the brain and central nervous system. Mutations in this gene can also cause autosomal dominant deafness 6 (DFNA6), also known as DFNA14 or DFNA38. Alternatively spliced transcript variants have been found for this gene. [provided by RefSeq, Mar 2009],
matters needing attention	Avoid repeated freezing and thawing!
Usage suggestions	This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

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