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ROGDI Rabbit pAb

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Catalog # AP57837

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9GZN7
Predicted	Rat, Pig, Human, Mouse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	32254
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human ROGDI
Epitope Specificity	31-130/287
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	Nucleus.
DISEASE	The disease is caused by mutations affecting the gene represented in this entry. Disease description:An autosomal recessive disorder characterized by severe global developmental delay, early-onset intractable seizures, spasticity, and amelogenesis imperfecta affecting both primary and secondary teeth and causing yellow or brown discoloration of the teeth. Although the phenotype is consistent, there is variability. Intellectual disability is related to the severity of seizures, and the disorder can thus be considered an epileptic encephalopathy. Some infants show normal development until seizure onset, whereas others are delayed from birth. The most severely affected individuals have profound mental retardation, never acquire speech, and become bedridden early in life.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a protein of unknown function. Loss-of-function mutation in this gene cause Kohlschutter-Tonz syndrome. Alternate splicing results in multiple transcript variants. [provided by RefSeq, Mar 2012]

Additional Information

Gene ID	79641
Other Names	Protein rogdi homolog, ROGDI
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	ROGDI {ECO:0000303|PubMed:22424600, ECO:0000312|HGNC:HGNC:29478}
Function	Probably involved in the regulation of intracellular pH (PubMed: 40049412). As part of the RAVE complex it may control the assembly of the proton-pumping V-ATPase complex and therefore regulate organelle acidification (Probable).
Cellular Location	Nucleus envelope. Presynapse {ECO:0000250|UniProtKB:Q4V7D2}. Cell projection, axon {ECO:0000250|UniProtKB:Q4V7D2}. Perikaryon {ECO:0000250|UniProtKB:Q4V7D2}. Cell projection, dendrite {ECO:0000250|UniProtKB:Q4V7D2}. Cytoplasmic vesicle, secretory vesicle, synaptic vesicle {ECO:0000250|UniProtKB:Q4V7D2}. Cytoplasm {ECO:0000250|UniProtKB:Q3TDK6}. Lysosome {ECO:0000250|UniProtKB:Q3TDK6}. Note=Detected primarily at presynaptic sites on axons, and to a lesser degree in soma and dendrites. Not detected at post-synaptic sites. {ECO:0000250|UniProtKB:Q4V7D2}
Tissue Location	Widely expressed with highest levels in spinal cord, brain, heart and bone marrow. Also expressed in fetal brain and liver.

Background

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