

NHLRC1 Rabbit pAb

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Catalog # AP59225

Product Information

Application	WB
Primary Accession	Q3SYB1
Reactivity	Rat
Predicted	Human, Mouse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human NHLRC1
Epitope Specificity	221-320/395
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. Nucleus. Localizes at the endoplasmic reticulum and, to a lesser extent, in the nucleus.
DISEASE	Defects in NHLRC1 are a cause of progressive myoclonic epilepsy type 2 (EPM2) [MIM:254780]; also known as Lafora disease. EPM2 is an autosomal recessive and severe form of adolescent-onset progressive epilepsy. Typically, as seizures increase in frequency, cognitive function declines towards dementia, and affected individuals die usually within 10 years after onset. EPM2 occurs worldwide, but it is particularly common in the mediterranean countries of southern Europe and northern Africa, in southern India and in the Middle East. At the cellular level, it is characterized by accumulation of starch-like polyglucosans called Lafora bodies (LBs) that are most abundant in organs with the highest glucose metabolism: brain, heart, liver and skeletal muscle. Among other conditions involving polyglucosans, EPM2 is unique in that the inclusions are in neuronal dendrites but not axons and the forming polyglucosan fibrils are associated with the endoplasmic reticulum.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Progressive myoclonic epilepsy type 2 (EPM2), also called Lafora disease, is an autosomal recessive disease characterized by grand mal seizures and/or myoclonus at about 15 years of age. Rapid and severe mental deterioration follows, often with psychotic features. Survival is less than 10 years after onset. Starch-like, endoplasmic reticulum-associated polyglucosans, called Lafora bodies, can be observed in brain, muscle, liver and heart. One cause of Lafora disease is due to mutations in NHLRC1, the gene encoding Malin. Forty-nine different mutations in NHLRC1 have been shown to cause EPM2. Malin, also called NHL repeat-containing protein 1, is a single subunit E3 ubiquitin ligase, containing 6 NHL repeats and 1 RING-type zinc finger. Malin's RING domain is responsible for its ability to mediate ubiquitination. Malin interacts with and polyubiquitinates Laforin, a protein also implicated in EPM2. Malin localizes to the endoplasmic reticulum and, to a lesser extent, in the nucleus. Malin is expressed in brain, cerebellum, spinal cord, medulla,

heart, liver, skeletal muscle and pancreas.

Additional Information

Dilution WB=1:500-2000

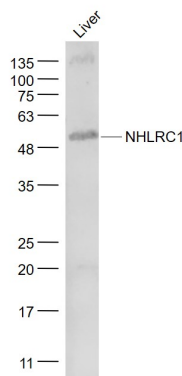
Storage Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Background

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Images



Sample:
Liver(Rat) Lysate at 40 ug
Primary: Anti- NHLRC1 (AP59225) at 1/1000 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 42 kD
Observed band size: 52 kD

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