Data Sheet (Cat.No.TMAB-09469)

Anti-NHLRC1 Polyclonal Antibody

Product Details

Ig Type:	IgG
Reactivity:	Rat (predicted:Human,Mouse,Rabbit)
Molecular Weight:	Theoretical: 42 kDa. Actual: 52 kDa.
Purification:	Protein A purified

Applications

Verified Activity:	Sample: liver (Rat) Lysate at 40 µg Primary: Anti-NHLRC1 (TMAB-09469) at 1/1000 dilution Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution Predicted band size: 42 kD Observed band size: 52 kD
Application:	WB
Recommended	WB: 1:500-2000

Properties

Stability & Storage:	Store at 2°C-8°C for 1 month. Store at -20°C or -80°C for 12 months. Avoid repeated freeze-thaw cycles.
Shipping:	Shipping with blue ice.

Antigen Details

Immunogen:	KLH conjugated synthetic peptide: human NHLRC1
Antigen Species:	Human
Gene ID:	378884
Uniprot ID:	Q3SYB1

Research Background

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.

Inhibitor · Natural Compounds · Compound Libraries · Recombinant Proteins

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