
NHLRC1 Antibody Blocking Peptide

Catalog Number: bs-9331P

Activity: Not tested

Purification: HPLC

Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.

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.