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Rabbit Anti-EFHC1 antibody

SL9013R

Product Name:	EFHC1
Chinese Name:	EFHC1蛋白抗体
Alias:	EF hand domain (C terminal) containing 1; EF hand domain containing protein 1; EF-hand domain-containing protein 1; Efhc1; EFHC1_HUMAN; EJA1; EJM1; FLJ10466; FLJ37290; JAE; Myoclonin 1; Myoclonin-1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:50-200（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	74kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human EFHC1:301-400/640
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	Defects in EFHC1 are the cause of juvenile myoclonic epilepsy type 1 (EJM1) [MIM:254770]. EJM1 is a subtype of idiopathic generalized epilepsy (IGE). Patients have afebrile seizures only, with onset in adolescence (rather than in childhood) and myoclonic jerks which usually occur after awakening and are triggered by sleep deprivation and fatigue.

Genetic variations in EFHC1 are the cause of susceptibility to juvenile absence epilepsy type 1 (JAE1) . JAE is a subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures.

Function:
May enhance calcium influx through CACNA1E and stimulate programmed cell death.

Subunit:
Interacts with the C-terminus of CACNA1E.

Tissue Specificity:
Widely expressed. Not detected in lymphocytes.

DISEASE:
Defects in EFHC1 are the cause of juvenile myoclonic epilepsy type 1 (EJM1) [MIM:254770]. EJM1 is a subtype of idiopathic generalized epilepsy (IGE). Patients have afebrile seizures only, with onset in adolescence (rather than in childhood) and myoclonic jerks which usually occur after awakening and are triggered by sleep deprivation and fatigue.
Genetic variations in EFHC1 are the cause of susceptibility to juvenile absence epilepsy type 1 (JAE1) [MIM:607631]. JAE is a subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures.

Similarity:
Contains 3 DM10 domains.
Contains 1 EF-hand domain.

SWISS:
Q5JVL4

Gene ID:
114327

Database links:

Entrez Gene: 114327 Human

Entrez Gene: 71877 Mouse

Omim: 608815 Human

SwissProt: Q5JVL4 Human

SwissProt: Q9D9T8 Mouse

	Unigene: 403171 Human Unigene: 29178 Mouse **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.