10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

EFHC1 Rabbit pAb

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Catalog # AP59103

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q5JVL4
Predicted	Rat, Pig, Dog, Human, Mouse, Chicken, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	73990
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human EFHC1
Epitope Specificity	301-400/640
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	114327
Other Names	EF-hand domain-containing protein 1, Myoclonin-1, EFHC1 (HGNC:16406)
Dilution	IHC-P=1:100-500,IHC-F=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	EFHC1 (HGNC:16406)
Function	Microtubule inner protein (MIP) part of the dynein-decorated doublet microtubules (DMTs) in cilia axoneme, which is required for motile cilia beating (PubMed:36191189). Microtubule-associated protein which regulates cell division and neuronal migration during cortical development (PubMed:19734894, PubMed:28370826). Necessary for radial and tangential cell migration during brain development, possibly acting as a regulator of cell morphology and process formation during migration (PubMed:22926142). May enhance calcium influx through CACNA1E and stimulate programmed cell death (PubMed:15258581, PubMed:19734894, PubMed:22926142, PubMed:28370826).
Cellular Location	Cytoplasm, cytoskeleton, cilium axoneme. Cytoplasm, cytoskeleton, flagellum axoneme {ECO:0000250|UniProtKB:Q9D9T8}. Cytoplasm, cytoskeleton, microtubule organizing center, centrosome. Cytoplasm, cytoskeleton, spindle. Cytoplasm, cytoskeleton, spindle pole
Tissue Location	Widely expressed. Not detected in lymphocytes.

Background

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