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GABA A Receptor gamma 2 Rabbit pAb

Catalog # AP99509

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	P18507
Other Accession	P18507
Host	Rabbit
Clonality	Polyclonal
Calculated MW	55186
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human GABRG2/GABA A Receptor gamma 2
Epitope Specificity	41-140/475
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the ligand-gated ion channel (TC 1.A.9) family. Gamma-aminobutyric acid receptor (TC 1.A.9.5) subfamily. GABRG2 sub-subfamily.
SUBUNIT	Generally pentameric. There are five types of GABA(A) receptor chains: alpha, beta, gamma, delta, and rho.
Post-translational modifications	Palmitoylated by ZDHHC3/GODZ; which may affect presynaptic clustering and/or cell surface stability.
DISEASE	Defects in GABRG2 are the cause of childhood absence epilepsy type 2 (ECA2) [MIM:607681]. ECA2 is a subtype of idiopathic generalized epilepsy (IGE) characterized by an onset at age 6-7 years, frequent absence seizures (several per day) and bilateral, synchronous, symmetric 3-Hz spike waves on EEG. During adolescence, tonic-clonic and myoclonic seizures develop. Some individuals manifest ECA2 occurring in combination with febrile convulsions. Defects in GABRG2 are the cause of familial febrile convulsions type 8 (FEB8) [MIM:611277]. A febrile convulsion is defined as a seizure event in infancy or childhood, usually occurring between 6 months and 6 years of age, associated with fever but without any evidence of intracranial infection or defined pathologic or traumatic cause. Febrile convulsions affect 5-12% of infants and children up to 6 years of age. There is epidemiological evidence that febrile seizures are associated with subsequent afebrile and unprovoked seizures in 2% to 7% of patients. Defects in GABRG2 are the cause of generalized epilepsy with febrile seizures plus type 3 (GEFS+3) [MIM:604233]. Generalized epilepsy with febrile seizures-plus refers to a rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. GEFS+ is a disease combining febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. Defects in GABRG2 are a cause of severe myoclonic epilepsy in infancy (SMEI) [MIM:607208]; also called Dravet syndrome. SMEI is a rare disorder characterized by generalized tonic, clonic, and tonic-clonic seizures that are initially induced by fever and begin during the first year of life. Later, patients

also manifest other seizure types, including absence, myoclonic, and simple and complex partial seizures. Psychomotor development delay is observed around the second year of life. SMEI is considered to be the most severe phenotype within the spectrum of generalized epilepsies with febrile seizures-plus.

Important Note This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Background Descriptions GABA, the major inhibitory neurotransmitter in the vertebrate brain, mediates neuronal inhibition by binding to the GABA/benzodiazepine receptor and opening an integral chloride channel.

Additional Information

Gene ID 2566

Other Names CAE2; DEE74; ECA2; EIEE74; FEB8; GEFSP3; GABAA-R; Gabrg-2; gamma2; GBRG2_BOVIN; GABRG2; GABA(A) receptor subunit gamma-2 (GABAAR subunit gamma-2); GBRG2_HUMAN; GBRG2_MOUSE; GBRG2_RAT; gamma-aminobutyric acid type A receptor subunit gamma2; gamma-aminobutyric acid (GABA) A receptor, gamma 2; GABA(A) receptor, gamma 2

Dilution WB=1:500-2000,IHC-P=1:200-800,IHC-F=1:200-800,IF=1:200-800

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 GABRG2 (HGNC:4087)

Function Gamma subunit of the heteropentameric ligand-gated chloride channel gated by gamma-aminobutyric acid (GABA), a major inhibitory neurotransmitter in the brain (PubMed:14993607, PubMed:16412217, PubMed:23909897, PubMed:2538761, PubMed:25489750, PubMed:27864268, PubMed:29950725, PubMed:30602789). GABA-gated chloride channels, also named GABA(A) receptors (GABAAR), consist of five subunits arranged around a central pore and contain GABA active binding site(s) located at the alpha and beta subunit interface(s) (PubMed:29950725, PubMed:30602789). When activated by GABA, GABAARs selectively allow the flow of chloride anions across the cell membrane down their electrochemical gradient (PubMed:14993607, PubMed:16412217, PubMed:2538761, PubMed:27864268, PubMed:29950725, PubMed:30602789). Gamma-2/GABRG2-containing GABAARs are found at both synaptic and extrasynaptic sites (By similarity). Chloride influx into the postsynaptic neuron following GABAAR opening decreases the neuron ability to generate a new action potential, thereby reducing nerve transmission (By similarity). GABAARs containing alpha-1 and beta-2 or -3 subunits exhibit synaptogenic activity; the gamma-2 subunit being necessary but not sufficient to induce rapid synaptic contacts formation (PubMed:23909897, PubMed:25489750). Extrasynaptic gamma-2- containing receptors contribute to the tonic GABAergic inhibition (By similarity). GABAARs function also as histamine receptor where histamine binds at the interface of two neighboring beta subunits and potentiates GABA response in a gamma-2 subunit-controlled manner (By similarity).

Cellular Location	Postsynaptic cell membrane; Multi-pass membrane protein {ECO:0000269|PubMed:30602789, ECO:0007744|PDB:6I53}. Cell membrane; Multi-pass membrane protein {ECO:0000269|PubMed:30602789, ECO:0007744|PDB:6I53} Cell projection, dendrite {ECO:0000250|UniProtKB:P22723}. Cytoplasmic vesicle membrane {ECO:0000250|UniProtKB:P18508}

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.