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CHRND Rabbit pAb

Catalog # AP98013

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	Q07001
Other Accession	Q07001
Predicted	Rat, Pig, Human, Mouse, Rabbit, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	58895
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human CHRND
Epitope Specificity	145-190/517
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. Acetylcholine receptor (TC 1.A.9.1) subfamily. Delta/CHRND sub-subfamily.
SUBUNIT	Pentamer of two alpha chains, and one each of the beta, delta, and gamma (in immature muscle) or epsilon (in mature muscle) chains.
DISEASE	Defects in CHRND are a cause of multiple pterygium syndrome lethal type (MUPSL) [MIM:253290]. Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. Defects in CHRND are a cause of congenital myasthenic syndrome slow-channel type (SCCMS) [MIM:601462]. SCCMS is the most common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. SCCMS is caused by kinetic abnormalities of the AChR, resulting in prolonged endplate currents and prolonged AChR channel opening episodes. Defects in CHRND are a cause of congenital myasthenic syndrome fast-channel type (FCCMS) [MIM:608930]. FCCMS is a congenital myasthenic syndrome characterized by kinetic abnormalities of the AChR. In most cases, FCCMS is due to mutations that decrease activity of the AChR by slowing the rate of opening of the receptor channel, speeding the rate of closure of the channel, or decreasing the number of openings of the channel during ACh occupancy. The result is failure to achieve threshold depolarization of the endplate and consequent failure to fire an action potential.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Members of the ligand-gated ion channel receptor family are characterized by

their fast transmitting response to neurotransmitters. Two important members of this family are the nicotinic acetylcholine and glutamate receptors, both of which are composed of five homologous subunits forming a transmembrane aqueous pore. These transmembrane receptors change conformation in response to their cognate neurotransmitter. Nicotinic acetylcholine receptors (AChRs) are found at the postsynaptic membrane of the neuromuscular junction and bind acetylcholine molecules, allowing ions to move through the pore. Glutamate receptors are found in the postsynaptic membrane of cells in the central nervous system. The activity that is generated at the synapse by the binding of acetylcholine is terminated by acetylcholinesterase, an enzyme that rapidly hydrolyzes acetylcholine. AChR delta, also known as CMS2A, FCCMS, SCCMS or CHRND, is a 517 amino acid multi-pass membrane protein that is associated with lethal type multiple pterygium syndrome, congenital myasthenic syndrome slow-channel type (SCCMS) and congenital myasthenic syndrome fast-channel type (FCCMS).

Additional Information

Gene ID	1144
Other Names	ACHRD; CMS2A; CMS3A; CMS3B; CMS3C; FCCMS; SCCMS; ACHD_HUMAN; CHRND; ACHD_RAT; Acrd; cholinergic receptor nicotinic delta subunit; cholinergic receptor, nicotinic, delta; cholinergic receptor, nicotinic, delta (muscle); cholinergic receptor, nicotinic delta; acetylcholine receptor, nicotinic, delta (muscle)
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=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	CHRND (HGNC:1965)
Synonyms	ACHRD
Function	After binding acetylcholine, the AChR responds by an extensive change in conformation that affects all subunits and leads to opening of an ion-conducting channel across the plasma membrane.
Cellular Location	Postsynaptic cell membrane; Multi-pass membrane protein. Cell membrane; Multi-pass membrane protein

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