

ACHG Rabbit pAb

CatalogNo: YN0391

Key Features

Host Species

- Rabbit

Reactivity

- Human, Mouse, Rat

Applications

- WB, ELISA

MW

- 56kD (Observed)

Isotype

- IgG

Storage

Storage* -15°C to -25°C/1 year (Do not lower than -25°C)

Formulation Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Recommended Dilution Ratios

WB 1:500-2000

ELISA 1:5000-20000

Basic Information

Clonality Polyclonal

Immunogen Information

Immunogen Synthesized peptide derived from human protein . at AA range: 30-110

Specificity ACHG Polyclonal Antibody detects endogenous levels of protein.

Target Information

Gene name CHRNG ACHRG

Protein Name Acetylcholine receptor subunit gamma

Organism	Gene ID	UniProt ID
Human	1146 ;	P07510 ;
Mouse		P04760 ;
Rat		P18916 ;

Cellular Localization Cell junction, synapse, postsynaptic cell membrane; Multi-pass membrane protein. Cell membrane; Multi-pass membrane protein.

Tissue specificity Muscle fibroblast,PCR rescued clones,Tongue,

Function Disease:Defects in CHRNG are a cause of Escobar syndrome [MIM:265000]; also called Escobar variant multiple pterygium syndrome or nonlethal type multiple pterygium syndrome. Escobar syndrome is a nonlethal form of arthrogyrosis multiplex congenita. It is an autosomal recessive condition characterized by excessive webbing (pterygia), congenital contractures (arthrogyrosis), and scoliosis. Variable other features include intrauterine death, congenital respiratory distress, short stature, faciocranial dysmorphism, ptosis, low-set ears, arachnodactyly and cryptorchism in males. Congenital contractures are common and may be caused by reduced fetal movements at sensitive times of development. Possible causes of decreased fetal mobility include space constraints such as oligohydramnion, drugs, metabolic conditions or neuromuscular disorders including myasthenia gravis.,Disease:Defects in CHRNG are a cause of lethal type multiple pterygium syndrome [MIM:253290]. Multiple pterygia are found infrequently in children with arthrogyrosis and in fetuses with fetal akinesia syndrome. Inheritance can be autosomal dominant, autosomal recessive, or X linked, but autosomal recessive inheritance appears to be most common. Clinical expression is very variable, and, in the severest form, lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia (e.g., chin to sternum, cervical, axillary, humero-ulnar, crural, popliteal, and ankles), and flexion contractures causing severe arthrogyrosis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies-in particular, cleft palate-are frequent.,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.,similarity:Belongs to the ligand-gated ionic channel (TC 1.A.9) family.,subunit:Pentamer of two alpha chains, and one each of the beta, delta, and gamma (in immature muscle) or epsilon (in mature muscle) chains.,

| Validation Data

| Contact information

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