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

SPG48 Rabbit pAb

Catalog # AP99636

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	O43299
Other Accession	O43299
Reactivity	Human
Host	Rabbit
Clonality	Polyclonal
Calculated MW	88605
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SPG48
Epitope Specificity	288-370/807
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBUNIT	Probably part of the adapter protein complex 5 (AP-5) atetramer composed of AP5B1, AP5M1, AP5S1 and AP5Z1. Interacts withZFYVE26 and SPG11.
DISEASE	Defects in AP5Z1 are the cause of spastic paraplegiaautosomal recessive type 48 (SPG48) [MIM:613647]. A form of spasticparaplegia, a neurodegenerative disorder characterized by a slow,gradual, progressive weakness and spasticity of the lower limbs.Rate of progression and the severity of symptoms are quitevariable. Initial symptoms may include difficulty with balance,weakness and stiffness in the legs, muscle spasms, and dragging thetoes when walking. In some forms of the disorder, bladder symptoms(such as incontinence) may appear, or the weakness and stiffnessmay spread to other parts of the body.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Chromosome 7 is about 158 milllion bases long, encodes over 1000 genes and makes up about 5% of the human genome. Chromosome 7 has been linked to Osteogenesis imperfecta, Pendred syndrome, Lissencephaly, Citrullinemia and Shwachman-Diamond syndrome. The deletion of a portion of the q arm of chromosome 7 is associated with Williams-Beuren syndrome, a condition characterized by mild mental retardation, an unusual comfort and friendliness with strangers and an elfin appearance. Deletions of portions of the q arm of chromosome 7 are also seen in a number of myeloid disorders including cases of acute myelogenous leukemia and myelodysplasia. KIAA0415 is a 807 amino acid protein that exists as three alternatively spliced isoforms. The KIAA0415 gene product has been provisionally designated KIAA0415 pending further characterization.

Additional Information

Gene ID	9907

Other Names	KIAA0415; SPG48; zeta; C330006K01Rik; AP5Z1_HUMAN; AP5Z1; Adaptor-related protein complex 5 zeta subunit (Zeta5); AP5Z1_MOUSE; adaptor related protein complex 5 subunit zeta 1; adaptor-related protein complex 5, zeta 1 subunit; adaptor related protein complex 5 zeta 1 subunit
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
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	AP5Z1
Synonyms	KIAA0415, SPG48
Function	As part of AP-5, a probable fifth adaptor protein complex it may be involved in endosomal transport. According to PubMed:20613862 it is a putative helicase required for efficient homologous recombination DNA double-strand break repair.
Cellular Location	Cytoplasm. Nucleus. Note=By SDS-PAGE, 2 isoforms have been observed, the shorter seems to be predominantly nuclear and the longer is mostly cytoplasmic.

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