SunLong Biotech Co.,LTD
Tel: 0086-571- 56623320 Fax:0086-571- 56623318
E-mail:sales@sunlongbiotech.com
www.sunlongbiotech.com

Rabbit Anti-SPG48 antibody

SL11760R

Product Name:	SPG48
Chinese Name:	SPG48蛋白抗体
Alias:	Hypothetical protein LOC9907; KIAA0415; Uncharacterized protein KIAA0415; AP5Z1_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	89kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SPG48:288-370/807
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	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.

Function:
The function of SPG48 remains unknown. There are 3 named isoforms produced by alternative splicing.

Subunit:
Probably part of the adapter protein complex 5 (AP-5) atetramer composed of AP5B1, AP5M1, AP5S1 and AP5Z1. Interacts withZFYVE26 and SPG11.

Subcellular Location:
Cytoplasm. Nucleus. Note=By SDS-PAGE, 2isoforms have been observed, the shorter seems to be predominantlynuclear and the longer is mostly cytoplasmic (PubMed:20613862).

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.

SWISS:
O43299

Gene ID:
9907

Database links:

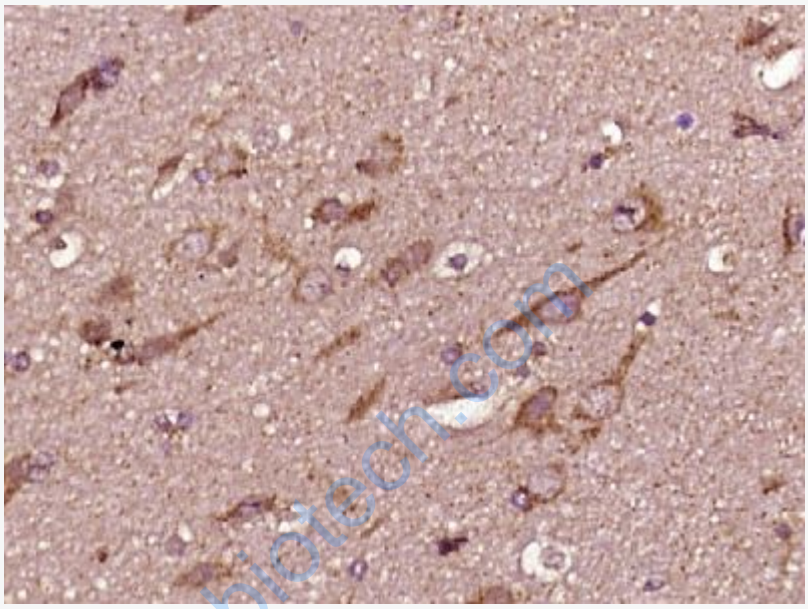
Entrez Gene: 9907Human

Entrez Gene: 231855Mouse

Omim: 613653Human

SwissProt: O43299Human

SwissProt: Q3U829Mouse

	Important Note: This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Picture:	 Paraformaldehyde-fixed, paraffin embedded (Human brain glioma); Antigen retrieval by boiling in sodium citrate buffer (pH6.0) for 15min; Block endogenous peroxidase by 3% hydrogen peroxide for 20 minutes; Blocking buffer (normal goat serum) at 37°C for 30min; Antibody incubation with (SPG48) Polyclonal Antibody, Unconjugated (SL11760R) at 1:400 overnight at 4°C, followed by operating according to SP Kit(Rabbit) (sp-0023) instructionsand DAB staining.