

Rabbit Anti-C9orf72 antibody

SL8595R

Product Name:	C9orf72
Chinese Name:	9号染色体开放阅读框72抗体
Alias:	chromosome 9 open reading frame 72; CI072_HUMAN; MGC23980; Uncharacterized protein C9orf72.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800Flow- Cyt=1ug/testICC=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:	53kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human C9orf72:391-481/481
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 9 consists of about 145 million bases and 4% of the human genome and encodes nearly 900 genes. Considered to play a role in gender determination, deletion of the distal portion of 9p can lead to development of male to female sex reversal, the phenotype of a female with a male X,Y genotype. Hereditary hemorrhagic telangiectasia, which is characterized by harmful vascular defects, is associated with the chromosome 9 gene encoding endoglin protein, ENG. Familial dysautonomia is also

associated with chromosome 9 though through the gene IKBKAP. Notably, chromosome 9 encompasses the largest interferon family gene cluster. Chromosome 9 is partnered with chromosome 22 in the translocation leading to the aberrant production of BCR-ABL fusion protein often found in leukemias. The C9orf72 gene product has been provisionally designated C9orf72 pending further characterization. There are two isoforms of C9orf72 that are produced as a result of alternative splicing events.

Subcellular Location:

Cytoplasm. Nucleus. Note=Detected in the cytoplasm of neurons from post mortem brain tissue (PubMed:21944778). Detected in the nucleus in fibroblasts (PubMed:21944779).

Tissue Specificity:

Both isoforms are widely expressed, including kidney, lung, liver, heart, testis and several brain regions, such as cerebellum. Also expressed in the frontal cortex and in lymphoblasts (at protein level).

DISEASE:

Defects in C9orf72 are the cause of frontotemporal dementia and/or amyotrophic lateral sclerosis (FTDALS) [MIM:105550]. An autosomal dominant neurodegenerative disorder characterized by adult onset of frontotemporal dementia and/or amyotrophic lateral sclerosis in an affected individual. There is high intrafamilial variation. Frontotemporal dementia is characterized by frontal and temporal lobe atrophy associated with neuronal loss, gliosis, and dementia. Patients exhibit progressive changes in social, behavioral, and/or language function. Amyotrophic lateral sclerosis is characterized by the death of motor neurons in the brain, brainstem, and spinal cord, resulting in fatal paralysis. Note=Caused by a large expansion of a GGGGCC hexanucleotide within the first C9orf72 intron located between the first and the second non-coding exons. The expansion leads to the loss of transcription of one of the two transcripts encoding isoform 1 and to the formation of nuclear RNA foci.

SWISS: Q96LT7

Gene ID: 203228

Database links:

Entrez Gene: 203228Human

Entrez Gene: 73205 Mouse

Entrez Gene: 313155Rat

<u>Omim: 614260</u>Human

SwissProt: Q96LT7Human
SwissProt: Q6DFW0Mouse
<u>SwissProt: Q66HC3</u> Rat
Unigene: 493639Human
Unigene: 331544Mouse
Unigene: 233897Rat
Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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