

Rabbit Anti-CT054 antibody

SL4164R

Product Name:	CT054
Chinese Name:	核黄素Transporter2抗体
Alias:	bA371L19.1; BVVLS; C20orf54; C20orf54provided by HGNC; Chromosome 20 open
	reading frame 54; hRF12; MGC10698; S52A3_HUMAN; RF12; Riboflavin transporter 2.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Cow,
Applications:	WB=1:500-2000ELISA=1:500-1000Flow-Cyt=3ug/Test
	not yet tested in other applications.
	optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	51kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human CT054/C20orf54:391-469/469
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:	CT054 is a Riboflavin transporter. Riboflavin transport is Na+-independent but
	moderately pH-sensitive. Activity is strongly inhibited by riboflavin analogs, such as
	lumiflavin, flavin mononucleotide (FMN) and flavin adenine dinucleotide (FAD), and
	to a lesser extent by amiloride.
	Function:

Riboflavin transporter. Riboflavin transport is Na(+)-independent but moderately pHsensitive. Activity is strongly inhibited by riboflavin analogs, such as lumiflavin, flavin mononucleotide (FMN) and flavin adenine dinucleotide (FAD), and to a lesser extent by amiloride.

Subcellular Location:

Cell membrane; Multi-pass membrane protein.

Tissue Specificity:

Predominantly expressed in testis. Highly expressed in small intestine and prostate.

DISEASE:

Defects in RFT2 are the cause of Brown-Vialetto-Van Laere syndrome (BVVLS) [MIM:211530]. BVVLS is rare autosomal recessive neurologic disorder characterized by sensorineural hearing loss and a variety of cranial nerve palsies, which develop over a relatively short period of time in a previously healthy individual. Sensorineural hearing loss may precede the neurological signs. The course is invariably progressive, but the rate of decline is variable within and between families. With disease evolution, long tract signs, lower motor neuron signs, cerebellar ataxia, and lower cranial nerve (III-VI) palsies develop, giving rise to a complex picture resembling amyotrophic lateral sclerosis. Diaphragmatic weakness and respiratory compromise are some of the most distressing features, leading to recurrent chest infections and respiratory failure, which are often the cause of patients' demise.

Defects in RFT2 are the cause of Fazio-Londe disease (FALOND) [MIM:211500]. A rare neurological disease characterized by progressive weakness of the muscles innervated by cranial nerves of the lower brain stem. It may present in childhood with severe neurological deterioration with hypotonia, respiratory insufficiency leading to premature death, or later in life with bulbar weakness which progresses to involve motor neurons throughout the neuroaxis. Clinical manifestations include dysarthria, dysphagia, facial weakness, tongue weakness, and fasciculations of the tongue and facial muscles.

Similarity:

Belongs to the riboflavin transporter family.

SWISS: Q9NQ40

Gene ID: 113278

Database links:

Entrez Gene: 113278 Human

SwissProt: Q9NQ40 Human



