

Rabbit Anti-Microcephalin 1/BRIT1 antibody

SL11227R

Product Name:	Microcephalin 1/BRIT1
Chinese Name:	小脑症基因1/认知相关蛋白抗体
Alias:	BRCT repeat inhibitor of TERT expression 1; BRIT 1; FLJ12847; Hypothetical protein FLJ12847; MCPH 1; MCPH1; MCPH1_HUMAN; MCT antibody Microcephalin 1; Microcephalin-1; Microcephaly primary autosomal recessive 1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Dog,
Applications:	WB=1:500-2000ELISA=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:	93kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human Microcephalin 1/BRIT1:11- 110/835
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:	Microcephalin modulates brain size and has been proliferating under strong positive selection for several thousand years, although the nature of the positive selection is poorly understood. Human Microcephalin contains three BRCA1 C-terminal (BRCT) domains and shares 57% identity with its mouse ortholog, the most conserved regions

being BRCT domains where there is 80% identity. Predominant expression of human Microcephalin is observed in fetal brain, liver and kidney tissues and is expressed during neurogenesis in mice. Microcephalin displays significantly higher rates of protein evolution in primates than in rodents; this trend is most noticeable for the subset of genes associated with nervous system development. Microcephalin has a very young, single nucleotide, polymorphism haplotype associated with modern humans; this gene is presumably still evolving in Homo sapiens. It functions in DNA damage response and regulation of cell cycle checkpoints.

Function:

Implicated in chromosome condensation and DNA damage induced cellular responses. May play a role in neurogenesis and regulation of the size of the cerebral cortex.

Subunit: Contains 3 BRCT domains.

Subcellular Location: Cytoplasm, cytoskeleton, centrosome.

Tissue Specificity: Expressed in fetal brain, liver and kidney.

DISEASE:

Defects in MCPH1 are the cause of microcephaly primary type 1 (MCPH1) [MIM:251200]; also known as true microcephaly or microcephaly vera. Microcephaly is defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits. This entity is inherited as autosomal recessive trait.

Similarity: Contains 3 BRCT domains.

SWISS: Q8NEM0

Gene ID: 79648

Database links:

Entrez Gene: 79648Human

Entrez Gene: 244329Mouse

	Omim: 607117Human
	SwissProt: Q8NEM0Human
	SwissProt: Q7TT79Mouse
	<u>Unigene: 656769</u> Human
	Unigene: 708770Human
	Unigene: 721952Human
	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 (mouse brain); 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 (Microcephalin 1, BRIT1) Polyclonal
	Antibody, Unconjugated (SL11227R) at 1:200 overnight at 4°C, followed by
	operating according to SP Kit(Rabbit) (sp-0023) instructions and DAB staining.

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