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Microcephalin 1/BRIT1 Rabbit pAb

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Catalog # AP54430

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q8NEM0
Predicted	Dog, Human, Mouse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	92849
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Microcephalin 1/BRIT1
Epitope Specificity	11-110/835
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cytoplasm, cytoskeleton, centrosome.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	79648

Other Names	Microcephalin, MCPH1 (HGNC:6954)
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	MCPH1 (HGNC:6954)
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.
Cellular Location	Cytoplasm, cytoskeleton, microtubule organizing center, centrosome
Tissue Location	Expressed in fetal brain, liver and kidney.

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.