

C16orf57 Rabbit pAb

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Catalog # AP59316

Product Information

Application	WB
Primary Accession	Q9BQ65
Reactivity	Rat
Predicted	Pig, Dog, Human, Mouse, Rabbit, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	30268
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human C16orf57
Epitope Specificity	171-265/265
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	Nucleus.
DISEASE	Defects in USB1 are the cause of poikiloderma with neutropenia (PN) [MIM:604173]. PN is a genodermatosis characterized by poikiloderma, pachyonychia and chronic neutropenia. The disorder starts as a papular erythematous rash on the limbs during the first year of life. It gradually spreads centripetally and, as the papular rash resolves, hypo- and hyperpigmentation result, with development of telangiectasias. Another skin manifestation is pachyonychia, but alopecia and leukoplakia are distinctively absent. One of the most important extracutaneous symptoms is an increased susceptibility to infections, mainly affecting the respiratory system, primarily due to a chronic neutropenia and to neutrophil functional defects. Bone marrow abnormalities account for neutropenia and may evolve into myelodysplasia associated with the risk of leukemic transformation. Poikiloderma with neutropenia shows phenotypic overlap with Rothmund-Thomson syndrome.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Involvement in disease;Defects in C16orf57 are the cause of poikiloderma with neutropenia (PN). PN is a genodermatosis characterized by poikiloderma, pachyonychia and chronic neutropenia. The disorder starts as a papular erythematous rash on the limbs during the first year of life. It gradually spreads centripetally and, as the papular rash resolves, hypo- and hyperpigmentation result, with development of telangiectasias. Another skin manifestation is pachyonychia, but alopecia and leukoplakia are distinctively absent. One of the most important extracutaneous symptoms is an increased susceptibility to infections, mainly affecting the respiratory system, primarily due to a chronic neutropenia and to neutrophil functional defects. Bone marrow abnormalities account for neutropenia and may evolve into myelodysplasia associated with the risk of leukemic transformation. Poikiloderma with neutropenia shows phenotypic overlap with

Additional Information

Gene ID	79650
Other Names	U6 snRNA phosphodiesterase 1, hUsb1, 3'-5' RNA exonuclease USB1, 4.6.1.-, Mutated in poikiloderma with neutropenia protein 1, Mutated in PN protein 1, hMpn1, USB1 {ECO:0000255 HAMAP-Rule:MF_03040, ECO:0000312 HGNC:HGNC:25792}
Dilution	WB=1:500-2000
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

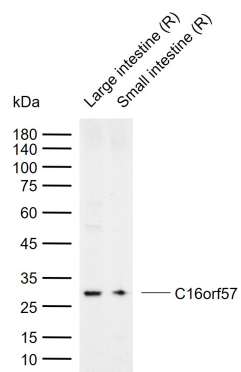
Name	USB1 {ECO:0000255 HAMAP-Rule:MF_03040, ECO:0000312 HGNC:HGNC:25792}
Function	3'-5' RNA exonuclease that trims the 3' end of oligo(U) and oligo(A) tracts of the pre-U6 small nuclear RNA (snRNA) molecule, leading to the formation of a mature U6 snRNA 3' end-terminated with a 2',3'-cyclic phosphate (PubMed: 22899009 , PubMed: 23022480 , PubMed: 23190533 , PubMed: 26213367 , PubMed: 28887445 , PubMed: 30215753 , PubMed: 31832688). Participates in the U6 snRNA 3' end processing that prevents U6 snRNA degradation (PubMed: 22899009 , PubMed: 23022480 , PubMed: 23190533 , PubMed: 26213367 , PubMed: 28887445 , PubMed: 30215753 , PubMed: 31832688). In addition also removes uridines from the 3' end of U6atac snRNA and possibly the vault RNA VTRNA1-1 (PubMed: 26213367).
Cellular Location	Nucleus {ECO:0000255 HAMAP-Rule:MF_03040, ECO:0000269 PubMed:22899009, ECO:0000269 PubMed:23022480}

Background

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Images

Sample:
 Lane 1: Rat Large intestine tissue lysates
 Lane 2: Rat Small intestine tissue lysates
 Primary: Anti-C16orf57 (AP59316) at 1/1000 dilution
 Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
 Predicted band size: 30 kDa
 Observed band size: 30 kDa



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