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Rabbit Anti-C16orf57 antibody

SL9631R

Product Name:	C16orf57
Chinese Name:	16号染色体开放阅读框57抗体
Alias:	Chromosome 16 open reading frame 57; CP057_HUMAN; FLJ13154; UPF0406 protein C16orf57.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Rabbit,Sheep,
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:	30kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human C16orf57:171-265/265
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:	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 Rothmund-Thomson syndrome.

Function:
Putative phosphodiesterase responsible for the U6 snRNA 3' end processing. Acts as a ribonuclease (RNase) responsible for trimming the poly(U) tract of the last nucleotides in the pre-U6 snRNA molecule, leading to the formation of mature U6 snRNA 3' end-terminated with a 2',3'-cyclic phosphate.

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.

Similarity:
Belongs to the USB1 family.

SWISS:
Q9BQ65

Gene ID:
79650

Database links:

Entrez Gene: 79650Human

SwissProt: Q9BQ65Human

Unigene: 408702Human

	Unigene: 588873Human **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.