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Rabbit Anti-DKC1 antibody

SL13007R

Product Name:	DKC1
Chinese Name:	核仁蛋白NAP57抗体
Alias:	CBF5; CBF5 homolog; Cbf5p homolog; DKC 1; DKC; Dkc1; DKC1_HUMAN; DKCX; Dyskeratosis congenita 1; Dyskeratosis congenita 1 dyskerin; Dyskerin; H/ACA ribonucleoprotein complex subunit 4; NAP 57; NAP57; NAP-57; NOLA 4; NOLA4; Nopp140 associated protein of 57 kDa; Nopp140-associated protein of 57 kDa; Nucleolar protein family A member 4; Nucleolar protein NAP57; snoRNP protein DKC1; XAP 101; XAP101.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,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:	58kDa
Cellular localization:	The nucleuscytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human DKC1/Dyskerin/NAP57:81-190/514
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:	Dyskerin (NAP57) associates with the chaperone protein Nopp140 and forms a small

ribonucleoprotein particle with GAR1 (NOLA1), NHP2 (NOLA2) and Nop10 for the isomerization of uridine to pseudouridine (1). GAR1, NHP2 and dyskerin localize to the dense fibrillar component of the nucleolus and in nuclear Cajal bodies (2). The dyskerin gene maps to chromosome Xq28 (3). Missense mutations in the dyskerin gene interfere with normal nuclear localization of dyskerin and cause Dyskeratosis congenita (DKC) (4). DKC is a rare, X-linked bone marrow disorder characterized by cutaneous hyperpigmentation, dystrophy of the nails, atrophy of the testicles and leukoplakia of the oral mucosa. The GAR1 gene maps to chromosome 4q25 (5,6). The NHP2 gene maps to chromosome 5q35.3 and encodes a 155-amino acid protein (2,7).

Function:
Isoform 1: Required for ribosome biogenesis and telomere maintenance. Probable catalytic subunit of H/ACA small nucleolar ribonucleoprotein (H/ACA snoRNP) complex, which catalyzes pseudouridylation of rRNA. This involves the isomerization of uridine such that the ribose is subsequently attached to C5, instead of the normal N1. Each rRNA can contain up to 100 pseudouridine ('psi') residues, which may serve to stabilize the conformation of rRNAs. Also required for correct processing or intranuclear trafficking of TERC, the RNA component of the telomerase reverse transcriptase (TERT) holoenzyme.

Subcellular Location:
Cytoplasm and Nucleus, nucleolus. Nucleus, Cajal body. Also localized to Cajal bodies.

Tissue Specificity:
Ubiquitously expressed.

DISEASE:
Defects in DKC1 are a cause of dyskeratosis congenita X-linked recessive (XDKC) [MIM:305000]. XDKC is a rare, progressive bone marrow failure syndrome characterized by the triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. Defects in DKC1 are the cause of Hoyeraal-Hreidarsson syndrome (HHS) [MIM:300240]. HHS is a multisystem disorder affecting males and is characterized by aplastic anemia, immunodeficiency, microcephaly, cerebellar hypoplasia, and growth retardation.

Similarity:
Belongs to the pseudouridine synthase TruB family.
Contains 1 PUA domain.

SWISS:
O60832

Gene ID:
1736

	Database links: Entrez Gene: 1736Human Entrez Gene: 245474Mouse Entrez Gene: 170944Rat Omim: 300126Human SwissProt: O60832Human SwissProt: Q9ESX5Mouse SwissProt: P40615Rat Unigene: 4747Human Unigene: 291062Mouse Unigene: 4223Rat **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.