

Product name:	Dyskerin Rabbit Polyclonal Antibody
Cat number:	ABN10244
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human Dyskerin. AA range:171-220
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:200-1:1000,ELISA 1:20000-1:40000
Molecular Weight:	57kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

dyskerin pseudouridine synthase 1(DKC1) Homo sapiens This gene functions in two distinct complexes. It plays an active role in telomerase stabilization and maintenance, as well as recognition of snoRNAs containing H/ACA sequences which provides stability during biogenesis and assembly into H/ACA small nucleolar RNA ribonucleoproteins (snoRNPs). This gene is highly conserved and widely expressed, and may play additional roles in nucleo-cytoplasmic shuttling, DNA damage response, and cell adhesion. Mutations have been associated with X-linked dyskeratosis congenita. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jan 2014],catalytic activity:RNA uridine = RNA pseudouridine.,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.,disease: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.,function: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.,online information:DKC1 mutation db,similarity:Belongs to the pseudouridine synthase truB family.,similarity:Contains 1 PUA domain.,subcellular location:Also localized to Cajal bodies (coiled bodies),subunit:Part of the H/ACA small nucleolar ribonucleoprotein (H/ACA snoRNP) complex, which contains NHP2/NOLA2, GAR1/NOLA1, NOP10/NOLA3, and DKC1/NOLA4, which is presumed to be the catalytic subunit. The complex contains a stable core formed by binding of one or two NOP10-DKC1 heterodimers to NHP2; GAR1 subsequently binds to this core via DKC1. The complex binds a box H/ACA small nucleolar RNA (snoRNA), which may target the specific site of modification within the RNA substrate. During assembly, the complex contains NAF1 instead of GAR1/NOLA1. The complex also interacts with TERC, which contains a 3'-terminal domain related to the box H/ACA snoRNAs. Specific interactions with snoRNAs or TERC are mediated by GAR1 and NHP2. Associates with NOLC1/NOPP140. H/ACA snoRNPs interact with the SMN complex, consisting of SMN1 or SMN2, SIP1/GEMIN2, DDX20/GEMIN3, and GEMIN4. This is mediated by interaction between GAR1 and SMN1 or SMN2. The SMN complex may be required for correct assembly of the H/ACA snoRNP complex. Component of the telomerase holoenzyme complex at least composed of TERT, DKC1, WDR79/TCAB1, NOP10, NHP2, GAR1, TEP1, EST1A, POT1 and a telomerase RNA template component (TERC),tissue specificity:Ubiquitously expressed.,