



Dyskerin Monoclonal Antibody

Catalog No	YP-mAb-01670
Isotype	IgG
Reactivity	Human;Mouse;Rat
Applications	WB
Gene Name	DKC1
Protein Name	H/ACA ribonucleoprotein complex subunit 4
Immunogen	The antiserum was produced against synthesized peptide derived from human Dyskerin. AA range:171-220
Specificity	Dyskerin Monoclonal Antibody detects endogenous levels of Dyskerin protein.
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-1:2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	DKC1; NOLA4; H/ACA ribonucleoprotein complex subunit 4; CBF5 homolog; Dyskerin; Nopp140-associated protein of 57 kDa; Nucleolar protein NAP57; Nucleolar protein family A member 4; snoRNP protein DKC1
Observed Band	57kD
Cell Pathway	[Isoform 1]: Nucleus, nucleolus . Nucleus, Cajal body . Also localized to Cajal bodies (coiled bodies). .; [Isoform 3]: Cytoplasm .
Tissue Specificity	Ubiquitously expressed.
Function	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.



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],

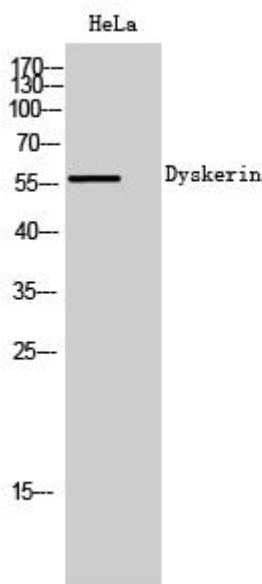
matters needing attention

Avoid repeated freezing and thawing!

Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

Products Images



Western Blot analysis of various cells using Dyskerin Monoclonal Antibody