

Dyskerin Polyclonal Antibody

Catalog No: YT1439

Reactivity: Human; Mouse; Rat

Applications: WB;IHC;IF;ELISA

Target: Dyskerin

Fields: >>Ribosome biogenesis in eukaryotes

O60832

Q9ESX5

Gene Name: DKC1

Protein Name: H/ACA ribonucleoprotein complex subunit 4

Human Gene ld: 1736

Human Swiss Prot

No:

Mouse Gene ld: 245474

Mouse Swiss Prot

No:

Rat Gene Id: 170944

Rat Swiss Prot No: P40615

Immunogen: The antiserum was produced against synthesized peptide derived from human

Dyskerin. AA range:171-220

Specificity: Dyskerin Polyclonal Antibody detects endogenous levels of Dyskerin protein.

Formulation : Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Source: Polyclonal, Rabbit, IgG

Dilution: WB 1:500 - 1:2000. IHC 1:100 - 1:300. IF 1:200 - 1:1000. ELISA: 1:40000. Not

yet tested in other applications.



Purification: The antibody was affinity-purified from rabbit antiserum by affinity-

chromatography using epitope-specific immunogen.

Concentration: 1 mg/ml

Storage Stability: -15°C to -25°C/1 year(Do not lower than -25°C)

Observed Band: 57kD

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],

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.

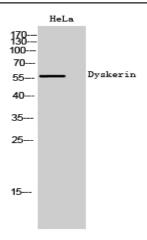
Subcellular Location:

[Isoform 1]: Nucleus, nucleolus . Nucleus, Cajal body . Also localized to Cajal

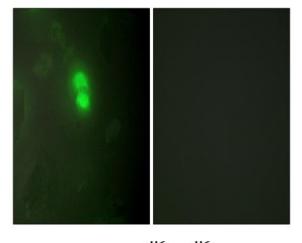
bodies (coiled bodies). .; [Isoform 3]: Cytoplasm .

Expression: Ubiquitously expressed.

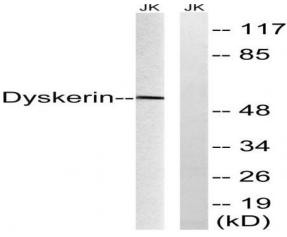
Products Images



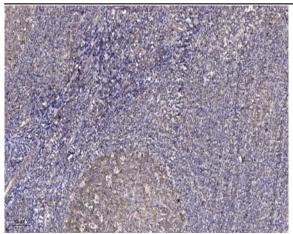
Western Blot analysis of HeLa cells using Dyskerin Polyclonal Antibody cells nucleus extracted by Minute TM Cytoplasmic and Nuclear Fractionation kit (SC-003,Inventbiotech,MN,USA).



Immunofluorescence analysis of HeLa cells, using Dyskerin Antibody. The picture on the right is blocked with the synthesized peptide.



Western blot analysis of lysates from JurKat cells, using Dyskerin Antibody. The lane on the right is blocked with the synthesized peptide.



Immunohistochemical analysis of paraffin-embedded human tonsil. 1, Antibody was diluted at 1:200(4° overnight). 2, Tris-EDTA,pH9.0 was used for antigen retrieval. 3,Secondary antibody was diluted at 1:200(room temperature, 45min).