abcam

Product datasheet

Recombinant Human DKC1/Dyskerin protein ab152335

1 Image

Description

Product name	Recombinant Human DKC1/Dyskerin protein
Expression system	Wheat germ
Accession	O60832
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	LRYEDGIEVNQEIVVITTKGEAICMAIALMTTAVISTCDHGIVAKIKRVI MERDTYPRKWGLGPKASQKKLMIKQGLLDKHGKPTDSTPATWKQEYVDYS
Predicted molecular weight	37 kDa including tags
Amino acids	321 to 420

Specifications

Our **Abpromise guarantee** covers the use of **ab152335** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA Western blot SDS-PAGE
Form	Liquid
Additional notes	This product was previously labelled as DKC1.

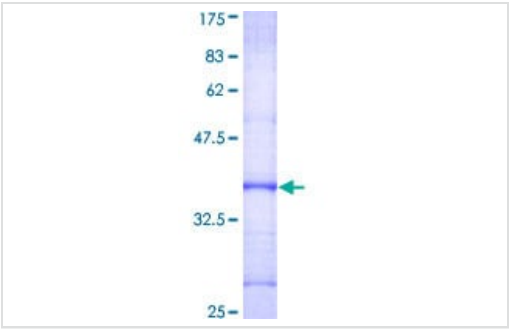
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

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. Isoform 3: Promotes cell to cell and cell to substratum adhesion, increases the cell proliferation rate and leads to cytokeratin hyper-expression (when overexpressed in HeLa cells).
Tissue specificity	Ubiquitously expressed.
Involvement in 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.
Sequence similarities	Belongs to the pseudouridine synthase TruB family. Contains 1 PUA domain.
Cellular localization	Cytoplasm and Nucleus > nucleolus. Nucleus > Cajal body. Also localized to Cajal bodies.

Images



SDS-PAGE - Recombinant Human DKC1/Dyskerin protein (ab152335)

12.5% SDS-PAGE analysis of ab152335 stained with Coomassie Blue.

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