abcam

Product datasheet

Recombinant Human DKC1/Dyskerin protein ab152335

1 图像

描述

产品名称	重组人DKC1/Dyskerin蛋白
表达系统	Wheat germ
Accession	O60832
蛋白长度	Protein fragment
无动物成分	No
性质	Recombinant
种属	Human
序列	LRYEDGIEVNQEIVVITTKGEAICMAIALMTTAVISTCDHGIVAKIKRVI MERDTYPRKWGLGPKASQKKLMIKQGLLDKHGKPTDSTPATWKQEYVDYS
预测分子量	37 kDa including tags
氨基酸	321 to 420

技术指标

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.

应用	ELISA Western blot SDS-PAGE
形式	Liquid
补充说明	This product was previously labelled as DKC1.

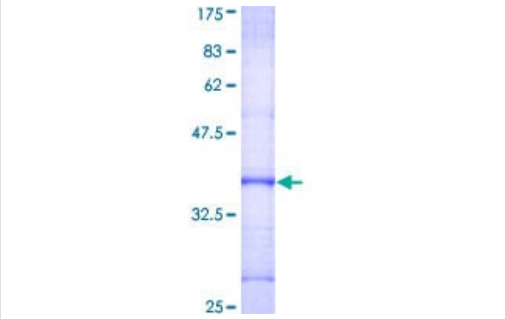
制备和贮存

稳定性和存储	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

常规信息

功能	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).
组织特异性	Ubiquitously expressed.
疾病相关	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.
序列相似性	Belongs to the pseudouridine synthase TruB family. Contains 1 PUA domain.
细胞定位	Cytoplasm and Nucleus > nucleolus. Nucleus > Cajal body. Also localized to Cajal bodies.

图片



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

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

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- We investigate all quality concerns to ensure our products perform to the highest standards

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