abcam

Product datasheet

Human DKC1 peptide ab71334

Overview

Product name Human DKC1 peptide

Description

Nature Synthetic

Specifications

Our Abpromise guarantee covers the use of **ab71334** in the following tested applications.

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

Purity 70 - 90% by HPLC.

Form Liquid

Additional notes

- First try to dissolve a small amount of peptide in either water or buffer. The more charged residues on a peptide, the more soluble it is in aqueous solutions.
- If the peptide doesn't dissolve try an organic solvent e.g. DMSO, then dilute using water or buffer.
- Consider that any solvent used must be compatible with your assay. If a peptide does not dissolve and you need to recover it, lyophilise to remove the solvent.
- Gentle warming and sonication can effectively aid peptide solubilisation. If the solution is cloudy or has gelled the peptide may be in suspension rather than solubilised.
- Peptides containing cysteine are easily oxidised, so should be prepared in solution just prior to use.

Preparation and Storage

Stability and Storage Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

Information available upon request.

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.

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