

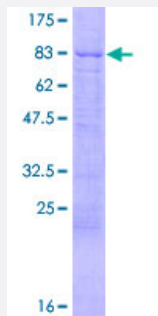
Full-Length

# DKC1 (Human) Recombinant Protein (P01)

Catalog # H00001736-P01

Size 25 ug, 10 ug

## Applications



## Specification

### Product Description

Human DKC1 full-length ORF ( NP\_001354.1, 1 a.a. - 514 a.a.) recombinant protein with GST-tag at N-terminal.

### Sequence

MADAEVILPKKHKKKKKERKSLPEEDVAEIQHAEFLIKPESKVAKLDTSQWPLLLKNFDKLNVRT  
THYTPLACGSNPLKREIGDYIRTGFINDKPSNPSSHEVVAWIRRLRVEKTGHSGTLDPKVTGCLV  
CIERATRLVKSQQSAGKEYVGVRLHNAIEGGTQLSRALETLTGALFQRPPPLAAVKRQLRVRTIYES  
KMIEYDPERRLGIFWVSCEAGTYIRTL CVHLGLLLGVGGQMQLRRVRSQVMSKDHMTMHDVL  
DAQWLYDNHKDESYLRRVVYPLEKLLTSHKRLVMKDSAVNAICYGAKIMLPGLRYEDGIEVNQEI  
VVITTKGEAICMAIALMTTAVISTCDHGVAKIKRVIMERDTPRKWGLGPKASQKKLMIKQGLLDKH  
GKPTDSTPATWKQEYVDYSESAKKEVVAEVVKAPQVVAEAAKTAKRKRESESESEDETTPAAPQ  
LIKKEKKKSKKDKKAKAGLESGAEPGDGSDTTKKKKKKKKAKEVELVSE

### Host

Wheat Germ (in vitro)

### Theoretical MW (kDa)

84.1

### Interspecies Antigen Sequence

Mouse (91); Rat (83)

### Preparation Method

[in vitro wheat germ expression system](#)

### Purification

Glutathione Sepharose 4 Fast Flow

### Quality Control Testing

12.5% SDS-PAGE Stained with Coomassie Blue.

Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
Storage Instruction	Store at -80°C. Aliquot to avoid repeated freezing and thawing.
Note	Best use within three months from the date of receipt of this protein.

## Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

## Gene Info — DKC1

Entrez GeneID	<a href="#">1736</a>
GeneBank Accession#	<a href="#">NM_001363.2</a>
Protein Accession#	<a href="#">NP_001354.1</a>
Gene Name	DKC1
Gene Alias	CBF5, DKC, FLJ97620, NAP57, NOLA4, XAP101
Gene Description	dyskeratosis congenita 1, dyskerin
Omim ID	<a href="#">300126</a> <a href="#">300240</a> <a href="#">305000</a>
Gene Ontology	<a href="#">Hyperlink</a>

## Gene Summary

This gene is a member of the H/ACA snoRNPs (small nucleolar ribonucleoproteins) gene family. snoRNPs are involved in various aspects of rRNA processing and modification and have been classified into two families: C/D and H/ACA. The H/ACA snoRNPs also include the NOLA1, 2 and 3 proteins. The protein encoded by this gene and the three NOLA proteins localize to the dense fibrillar components of nucleoli and to coiled (Cajal) bodies in the nucleus. Both 18S rRNA production and rRNA pseudouridylation are impaired if any one of the four proteins is depleted. These four H/ACA snoRNP proteins are also components of the telomerase complex. The protein encoded by this gene is related to the *Saccharomyces cerevisiae* Cbf5p and *Drosophila melanogaster* Nopp60B proteins. The gene lies in a tail-to-tail orientation with the palmitoylated erythrocyte membrane protein gene and is transcribed in a telomere to centromere direction. Both nucleotide substitutions and single trinucleotide repeat polymorphisms have been found in this gene. Mutations in this gene cause X-linked dyskeratosis congenita, a disease resulting in reticulate skin pigmentation, mucosal leukoplakia, nail dystrophy, and progressive bone marrow failure in most cases. Mutations in this gene also cause Hoyeraal-Hreidarsson syndrome, which is a more severe form of dyskeratosis congenita. Two transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq]

## Other Designations

H/ACA ribonucleoprotein complex subunit 4|OTTHUMP00000026046|cbf5p homolog|dyskerin|nopp140-associated protein of 57 kDa|nucleolar protein family A member 4|snoRNP protein DKC1

## Disease

- [Anemia](#)