



Recombinant Human H/ACA ribonucleoprotein complex subunit 4 (DKC1)

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| Product Code | CSB-EP006919HU |
| Storage | Store at -20°C, for extended storage, conserve at -20°C or -80°C. |
| Uniprot No. | O60832 |
| Product Type | Recombinant Protein |
| Immunogen Species | Homo sapiens (Human) |
| Purity | ≥85% (SDS-PAGE) |
| Sequence | <p>ADAEVIILP KKHKKKKERK SLPEEDVAEI QHAEFLIKP ESKVAKLDTS QWPLLLKNFD KLNVRTTHYT PLACGSNPLK REIGDYIRTG FINLDKPSNP SSHEVVAVIR RILRVEKTGH SGTLDPKVTG CLIVCIERAT RLVKSQQSAG KEYVGIVRLH NAIEGGTQLS RALETLTGAL FQRPPLIAAV KRQLRVRTIY ESKMIEYDPE RRLGIFWVSC EAGTYIRTLV VHLGLLLGVG GQMQLRRVR SGVMSEKDHM VTMHDVLDAQ WLYDNHKDES YLRRVVYPLE KLLTSHKRLV MKDSAVNAIC YGAKIMLPV LRYEDGIEVN QEIVVITTKG EAICMAIALM TTAVISTCDH GIVAKIKRVI MERDTYPRKW GLGPKASQKK LMIKQGLLDK HGKPTDSTPA TWKQEYVDYS ESAKKEVVAE VVKAPQVVAE AAKTAKRKRE SESEDETTP AAPQLIKKEK KKSCKDKKAK AGLESGAEPG DGSDTTKKK KKKKKAKEVE LVSE</p> |
| Source | E.coli |
| Target Names | DKC1 |
| Protein Names | Recommended name: H/ACA ribonucleoprotein complex subunit 4 EC= 5.4.99.- Alternative name(s): CBF5 homolog Dyskerin Nopp140-associated protein of 57 kDa Nucleolar protein NAP57 Nucleolar protein family A member 4 snoRNP |
| Expression Region | 2-514 |
| Notes | Repeated freezing and thawing is not recommended. Store working aliquots at 4°C for up to one week. |
| Tag Info | Tag type will be determined during the manufacturing process. |
| Protein Length | Full Length of Mature Protein |
| Target Details | <p>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. This protein 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. This protein is related to the <i>Saccharomyces cerevisiae</i> Cbf5p and <i>Drosophila melanogaster</i> Nop60B proteins. The gene lies in a tail-to-tail orientation with the palmitoylated erythrocyte membrane protein</p> |



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.

Reconstitution

We recommend that this vial be briefly centrifuged prior to opening to bring the contents to the bottom. Please reconstitute protein in deionized sterile water to a concentration of 0.1-1.0 mg/mL. We recommend to add 5-50% of glycerol (final concentration) and aliquot for long-term storage at -20°C/-80°C. Our default final concentration of glycerol is 50%. Customers could use it as reference.

Shelf Life

The shelf life is related to many factors, storage state, buffer ingredients, storage temperature and the stability of the protein itself. Generally, the shelf life of liquid form is 6 months at -20°C/-80°C. The shelf life of lyophilized form is 12 months at -20°C/-80°C.