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DKC1

Reactivity: Human Mouse Rat

Tested applications:WB IHC IF

Recommended Dilution:WB 1:500 - 1:2000 IHC 1:50 - 1:200 IF 1:50 - 1:200

Calculated MW:58kDa

Observed MW: Refer to Figures

Immunogen:

Recombinant protein of human DKC1

Storage Buffer:

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Concentration:

j

Synonym:

DKC1; DKC; CBF5; DKCX; NAP57; NOLA4; XAP101;

Catalog #:A1862

Antibody Type:

Polyclonal Antibody

Species: Rabbit

Gene ID:1736

Isotype: IgG

Swiss Prot:O60832

Purity: Affinity purification

For research use only.

Background:

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 Nop60B 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, Dec 2008]

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