

## Product datasheet for **TA323896**

### DKC1 Rabbit Polyclonal Antibody

#### Product data:

Product Type:	Primary Antibodies
Applications:	WB
Recommended Dilution:	WB: 1:500-2000
Reactivity:	Human, Mouse, Rat
Host:	Rabbit
Isotype:	IgG
Clonality:	Polyclonal
Immunogen:	Fusion protein corresponding to C terminal 270 amino acids of human dyskeratosis congenita 1, dyskerin
Formulation:	PBS pH7.3, 0.05% NaN <sub>3</sub> , 50% glycerol
Concentration:	lot specific
Purification:	Antigen affinity purification
Conjugation:	Unconjugated
Storage:	Store at -20°C as received.
Stability:	Stable for 12 months from date of receipt.
Predicted Protein Size:	58 kDa
Gene Name:	dyskerin pseudouridine synthase 1
Database Link:	<a href="#">NP_001135935</a> <a href="#">Entrez Gene 170944 Rat</a> <a href="#">Entrez Gene 245474 Mouse</a> <a href="#">Entrez Gene 1736 Human</a> <a href="#">O60832</a>

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**Background:**

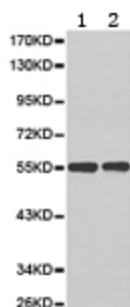
This gene is a member of the H/ACA snoRNPs (small nucleolar ribonucleoproteins) gene family. snoRNPs are involved in various aspects of rRNA processing and modification and have been classified into two families: C/D and H/ACA. The H/ACA snoRNPs also include the NOLA1, 2 and 3 proteins. The protein encoded 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.

**Synonyms:**

CBF5; DKC; DKCX; NAP57; NOLA4; XAP101

**Protein Families:**

Druggable Genome

**Product images:**


Predicted band size: 58 kDa. Positive control: Hela and MCF7 cell lysate. Recommended dilution: 1/500-2000. (Gel: 10%SDS-PAGE Lane 1: Hela cell lysate Lane 2: MCF7 cell lysate Lysate: 40 ug per lane Primary antibody: 1/500 dilution Secondary antibody: Goat anti Rabbit IgG - H&L (HRP) at 1/10000 dilution Exposure time: 1 minute)