

**Goat Anti-DKC1 Antibody**  
**Peptide-affinity purified goat antibody**  
**Catalog # AF1322a****Specification**

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**Goat Anti-DKC1 Antibody - Product Information**

Application	IHC, E
Primary Accession	<a href="#">O60832</a>
Other Accession	<a href="#">NP_001354</a> , <a href="#">1736</a> , <a href="#">245474 (mouse)</a>
Reactivity	Human
Predicted	Mouse
Host	Goat
Clonality	Polyclonal
Concentration	100ug/200ul
Isotype	IgG
Calculated MW	57674

**Goat Anti-DKC1 Antibody - Additional Information****Gene ID** 1736**Other Names**

H/ACA ribonucleoprotein complex subunit 4, 5.4.99.-, CBF5 homolog, Dyskerin, Nopp140-associated protein of 57 kDa, Nucleolar protein NAP57, Nucleolar protein family A member 4, snoRNP protein DKC1, DKC1, NOLA4

**Dilution**

IHC~~1:100~500

E~~N/A

**Format**

0.5 mg IgG/ml in Tris saline (20mM Tris pH7.3, 150mM NaCl), 0.02% sodium azide, with 0.5% bovine serum albumin

**Storage**

Maintain refrigerated at 2-8°C for up to 6 months. For long term storage store at -20°C in small aliquots to prevent freeze-thaw cycles.

**Precautions**

Goat Anti-DKC1 Antibody is for research use only and not for use in diagnostic or therapeutic procedures.

**Goat Anti-DKC1 Antibody - Protein Information****Name** DKC1 ([HGNC:2890](#))**Synonyms** NOLA4

### Function

[Isoform 1]: Catalytic subunit of H/ACA small nucleolar ribonucleoprotein (H/ACA snoRNP) complex, which catalyzes pseudouridylation of rRNA (PubMed:<a href="http://www.uniprot.org/citations/25219674" target="\_blank">25219674</a>, PubMed:<a href="http://www.uniprot.org/citations/32554502" target="\_blank">32554502</a>). This involves the isomerization of uridine such that the ribose is subsequently attached to C5, instead of the normal N1 (PubMed:<a href="http://www.uniprot.org/citations/25219674" target="\_blank">25219674</a>). Each rRNA can contain up to 100 pseudouridine ('psi') residues, which may serve to stabilize the conformation of rRNAs. Required for ribosome biogenesis and telomere maintenance (PubMed:<a href="http://www.uniprot.org/citations/19179534" target="\_blank">19179534</a>, PubMed:<a href="http://www.uniprot.org/citations/25219674" target="\_blank">25219674</a>). Also required for correct processing or intranuclear trafficking of TERC, the RNA component of the telomerase reverse transcriptase (TERT) holoenzyme (PubMed:<a href="http://www.uniprot.org/citations/19179534" target="\_blank">19179534</a>).

### Cellular Location

[Isoform 1]: Nucleus, nucleolus. Nucleus, Cajal body {ECO:0000250|UniProtKB:P40615}

### Tissue Location

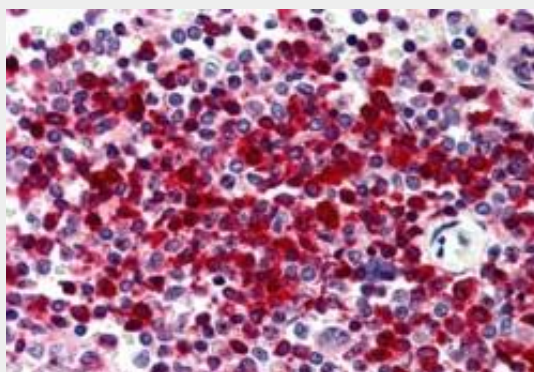
Ubiquitously expressed.

### Goat Anti-DKC1 Antibody - Protocols

Provided below are standard protocols that you may find useful for product applications.

- [Western Blot](#)
- [Blocking Peptides](#)
- [Dot Blot](#)
- [Immunohistochemistry](#)
- [Immunofluorescence](#)
- [Immunoprecipitation](#)
- [Flow Cytometry](#)
- [Cell Culture](#)

### Goat Anti-DKC1 Antibody - Images



AF1322a (5 µg/ml) staining of paraffin embedded Human Spleen. Steamed antigen retrieval with citrate buffer pH 6, AP-staining.

### Goat Anti-DKC1 Antibody - 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.

### **Goat Anti-DKC1 Antibody - References**

Novel dyskerin-mediated mechanism of p53 inactivation through defective mRNA translation. Montanaro L, et al. *Cancer Res*, 2010 Jun 1. PMID 20501855.

Inactivation of the tumor suppressor genes causing the hereditary syndromes predisposing to head and neck cancer via promoter hypermethylation in sporadic head and neck cancers. Smith IM, et al. *ORL J Otorhinolaryngol Relat Spec*, 2010. PMID 20332657.

Effects of dyskeratosis congenita mutations in dyskerin, NHP2 and NOP10 on assembly of H/ACA pre-RNPs. Trahan C, et al. *Hum Mol Genet*, 2010 Mar 1. PMID 20008900.

Novel mutations of the DKC1 gene in individuals affected with dyskeratosis congenita. Rostamiani K, et al. *Blood Cells Mol Dis*, 2010 Mar-Apr. PMID 19879169.

Single-molecule analysis of the human telomerase RNA-dyskerin interaction and the effect of dyskeratosis congenita mutations. Ashbridge B, et al. *Biochemistry*, 2009 Nov 24. PMID 19835419.