DKC1 (Human) Cell-Based ELISA Kit

Catalog # KA6314 Size 1 Kit

Specification

Product Description	DKC1 (Human) Cell-Based ELISA Kit is an indirect enzyme-linked immunoassay for qualitative determination of DKC1 expression in cultured cells.
Suitable Sample	Attached Cell, Loosely Attached Cell, Suspension Cell.
Label	HRP-conjugated
Detection Method	Colorimetric
Assay Type	Qualitative
Reactivity	Human, Mouse, Rat
Regulatory Status	For research use only (RUO)
Storage Instruction	Store at 4°C for six months.

Applications

- Qualitative

Gene Info — DKC1

Entrez GeneID	1736
Protein Accession#	O60832
Gene Name	DKC1
Gene Alias	CBF5, DKC, FLJ97620, NAP57, NOLA4, XAP101
Gene Description	dyskeratosis congenita 1, dyskerin

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Omim ID	300126 300240 305000
Gene Ontology	Hyperlink
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 cl assified 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 fi brillar components of nucleoli and to coiled (Cajal) bodies in the nucleus. Both 18S rRNA producti on and rRNA pseudouridylation are impaired if any one of the four proteins is depleted. These fou r 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 No p60B proteins. The gene lies in a tail-to-tail orientation with the palmitoylated erythrocyte membra ne protein gene and is transcribed in a telomere to centromere direction. Both nucleotide substitut ions 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. Mutatio ns in this gene also cause Hoyeraal-Hreidarsson syndrome, which is a more severe form of dyske ratosis congenita. Two transcript variants encoding different isoforms have been found for this ge ne. [provided by RefSeq
Other Designations	H/ACA ribonucleoprotein complex subunit 4|OTTHUMP00000026046|cbf5p homolog|dyskerin|no pp140-associated protein of 57 kDa|nucleolar protein family A member 4|snoRNP protein DKC1

Disease

- Anemia

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