DKC1 (dyskeratosis congenita 1, dyskerin)

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Published in Atlas Database: November 2002
Online updated version: http://AtlasGeneticsOncology.org/Genes/DKC1ID157.html
DOI: 10.4267/2042/37924

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Identity

HGNC (Hugo): DKC1
Location: Xq28
Local order: Distal, DKC1 is between DXS1684 and DXS1108.

Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

Note: X-linked dyskeratosis congenita (genes for dominant and recessive autosomal forms have not been identified).

DNA/RNA

Description
Gene composed of 15 exons (exons 1 and 15 non-coding) / 15 kb length.
cDNA 2465 bp (open reading frame between nt 93 and 1637).

Protein

Description
Dyskerin, 514 amino acids, 57 kDa.

Expression
Widespread tissue expression.

Function
Multifunctional nucleolar protein which associates with H+ACA (hairpin-linge hairpin-tail) class of small nucleolar RNA as its catalytic sub-unit; implicated in centromere function; associated also with the telomerase RNA component; function in ribosome biosynthesis.

Homology
Highly conserved in eukaryotes: Nap57 (nucleolar associated protein) in the rat, Nop60B in drosophila, Cbf5p (centromere/microtubule binding protein) in yeast. Regional homologies with bacterial Trub proteins and Saccharomyces cerevisiae PUS4 protein.

Implicated in

Disease
Dyskeratosis congenita, X-linked recessive form.

Hybrid/Mutated gene
Missense mutation by single-nucleotide substitution at position 1058 in exon 11 (A353V) detected in several different families. Sporadic other missense mutations were detected in exon 3, 4, 10, 12 and in intron 2. Rare deletions and no null mutations are observed.
Abnormal protein
Non functional protein. It is not presently known how the different mutations affect the protein activity and are responsible of the various phenotypes.

Oncogenesis
Myelodysplasia and leukemia following bone marrow failure and pancytopenia. Spinocellular carcinoma, other carcinomas of various localization.

References


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