

# Dyskeratosis Congenita

[View the current Dyskeratosis Congenita InDepth page on BioPortfolio.com \(PDF\)](#)

([http://www.bioportfolio.com/indepth/Dyskeratosis\\_Congenita.html](http://www.bioportfolio.com/indepth/Dyskeratosis_Congenita.html)) - Regularly Updated.

## Recent Publications on Dyskeratosis Congenita:



- [Dyskerin ablation in mouse liver inhibits ribosomal RNA processing and cell division.](#)  
*Dyskerin is a component of small nucleolar ribonucleoprotein complexes and...*18th November, 2009  
Division of Hematology, Department of Medicine, Department of Pediatrics,- Mol Cell Biol. 2009 Nov 16. ([DOI Direct Link](#))
- [Anomalous electrophoretic migration of newly synthesized ribosomal RNAs and their precursors from cells with DKC1 mutations.](#)  
*Mutations in the X-linked gene, DKC1, encoding dyskerin, cause...*6th November, 2009  
Division of Hematology, Department of Medicine, Washington University- FEBS Lett. 2009 Sep 17;583(18):3086-90. Epub 2009 Sep 2. ([DOI Direct Link](#))
- [Novel mutations of the DKC1 gene in individuals affected with dyskeratosis congenita.](#)  
*Dyskerin is a component of small nucleolar ribonucleoprotein complexes and...*3rd November, 2009  
Department of Biology, California State University Northridge, Northridge,- Blood Cells Mol Dis. 2009 Oct 28. ([DOI Direct Link](#))
- [DKC1 overexpression associated with prostate cancer progression.](#)  
*BACKGROUND: Dyskerin encoded by the DKC1 gene is a predominantly nucleolar...*29th October, 2009  
Urologische Klinik, Heinrich Heine Universitat, Dusseldorf, Germany.- Br J Cancer. 2009 Oct 20;101(8):1410-6. Epub 2009 Sep 15. ([DOI Direct Link](#))
- [Hoyeraal-Hreidarsson syndrome: magnetic resonance imaging findings.](#)  
*Hoyeraal-Hreidarsson syndrome (HH) has been defined as a severe variant of...*27th October, 2009  
Department of Radiology, Dokkyo Medical University, 880 Kitakobayashi,- Jpn J Radiol. 2009 Oct;27(8):324-7. Epub 2009 Oct 27. ([DOI Direct Link](#))
- [Retinal vasculopathy in a family with autosomal dominant dyskeratosis congenita.](#)  
*BACKGROUND: Dyskeratosis congenita is a rare multisystem bone marrow...*27th October, 2009  
Broad Institute, Cambridge, Massachusetts, USA.- Ophthalmic Genet. 2009 Dec;30(4):181-4.
- [Telomerase reverse transcriptase-dependent telomere equilibration mitigates tissue dysfunction in mTert heterozygotes.](#)  
*Autosomal dominant mutations in telomere-associated factors elicit a...*21st October, 2009  
Ontario Cancer Institute/Campbell Family Institute for Cancer Research,- Dis Model Mech. 2009 Nov-Dec;2(11-12):620-6. Epub 2009 Oct 19. ([DOI Direct Link](#))
- [Single-molecule analysis of the human telomerase RNA.dyskerin interaction and the effect of dyskeratosis congenita mutations.](#)  
*It has been proposed that human telomerase RNA (hTR) interacts with...*20th October, 2009

University Chemical Laboratories, University of Cambridge, Lensfield Road,- Biochemistry. 2009 Nov 24;48(46):10858-65. ([DOI Direct Link](#))

- [Fatal bilateral pneumothoraces complicating dyskeratosis congenita: a case report.](#)  
*INTRODUCTION: Dyskeratosis congenita is a rare genodermatosis,...*16th October, 2009  
- J Med Case Reports. 2009 Mar 26;3:6622. ([DOI Direct Link](#))
- [\[Mutation in the telomerase complex component NHP2 in dyskeratosis congenita\]](#)  
*INTRODUCTION: Dyskeratosis congenita is a rare genodermatosis,...*6th October, 2009  
Service de dermatologie, hopital Saint-Eloi, CHRU de Montpellier, 80,- Ann Dermatol Venereol. 2009 Oct;136(10):752-3. Epub 2009 Mar 17. ([DOI Direct Link](#))
- [Acquired monosomy 7 myelodysplastic syndrome in a child with clinical features suggestive of dyskeratosis congenita and IMAGE association.](#)  
*We describe a case of acquired monosomy 7 myelodysplastic syndrome (MDS)...*18th September, 2009  
Department of Pediatrics, Washington University School of Medicine, St.- *Pediatr Blood Cancer.* 2010 Jan;54(1):154-7. ([DOI Direct Link](#))
- [Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia.](#)  
*The primary pathology in many cases of myelodysplasia (MDS) and acute...*18th September, 2009  
Barts and The London School of Medicine and Dentistry, Queen Mary- *Hum Mutat.* 2009 Nov;30(11):1567-73. ([DOI Direct Link](#))
- [Pathogenic NAP57 mutations decrease ribonucleoprotein assembly in dyskeratosis congenita.](#)  
*X-linked dyskeratosis congenita (DC) is a rare bone marrow failure...*8th September, 2009  
Department of Anatomy and Structural Biology, Albert Einstein College of- *Hum Mol Genet.* 2009 Dec 1;18(23):4546-51. Epub 2009 Sep 4. ([DOI Direct Link](#))
- [Erosion of telomeric single-stranded overhang in patients with aplastic anaemia carrying telomerase complex mutations.](#)  
*BACKGROUND: Loss-of-function mutations in telomerase complex genes reduce...*14th August, 2009  
National Heart, Lung, and Blood Institute, National Institutes of Health,- *Eur J Clin Invest.* 2009 Nov;39(11):1025-32. Epub 2009 Aug 11. ([DOI Direct Link](#))
- [Telomere dysfunction in human diseases: the long and short of it!](#)  
*It has been over one hundred years since the first reported case of...*29th July, 2009  
Department of Pathology and Laboratory Medicine, Emory University Atlanta,- *Int J Clin Exp Pathol.* 2009 May 10;2(6):528-43.

## BioNews Results for Dyskeratosis Congenita

- [Mice Regain Ability To Extend Telomeres Suggesting Potential For Dyskeratosis Congenita Therapy](#)  
*Medical News Today: Oct 19 2009 4:05PM Matching: dyskeratosis congenita*
- [Medical News: Mice Regain Ability To Extend Telomeres Suggesting Potential For Dyskeratosis Congenita Therapy](#)  
*Medical News Today: Oct 19 2009 1:45PM Matching: dyskeratosis congenita*



## Dyskeratosis Congenita Patents:

- 5004681- [Preservation of fetal and neonatal hematopoietic stem and progenitor cells of the blood](#)
- 6555374- [Multiple mesodermal lineage differentiation potentials for adipose tissue-derived stromal cells and uses thereof](#)
- 6564120- [Storage system, particularly with automatic insertion and retrieval](#)
- 6569427- [Isolation and preservation of fetal and neonatal hematopoietic stem and progenitor cells of the blood](#)
- 6583109- [Therapeutic polypeptides from .beta.-hCG and derivatives](#)
- 6596688- [Method for promoting hematopoiesis](#)
- 6605275- [Isolation and preservation of fetal and neonatal hematopoietic stem and progenitor cells of the blood](#)
- 6620416- [Method for treating HIV](#)
- 6699834- [Method for treating cancer](#)
- 6759215- [Method of preparing human stem cell factor polypeptide](#)
- 6805882- [Therapeutic fractions of sources of HCG](#)
- 6461645- [Isolation and preservation of fetal and neonatal hematopoietic stem and progenitor cells of the blood](#)
- 6342358- [Human telomerase RNA elements](#)
- 5192553- [Isolation and preservation of fetal and neonatal hematopoietic stem and progenitor cells of the blood and methods of therapeutic use](#)
- 5401835- [Human erythroid p55 nucleic acids](#)
- 5968513- [Method of promoting hematopoiesis using derivatives of human chorionic gonadotropin](#)
- 6093699- [Method for gene therapy involving suppression of an immune response](#)
- 6204363- [Stem cell factor](#)
- 6207417- [DNA encoding stem cell factor](#)
- 6207454- [Method for enhancing the efficiency of gene transfer with stem cell factor \(SCF\) polypeptide](#)
- 6207802- [Stem cell factor and compositions](#)
- 6218148- [DNS encoding stem cell factor](#)
- 6248319- [Method for increasing hematopoietic progenitor cells by stem cell factor polypeptides](#)
- 6841147- [Stem cell factor compositions](#)
- 6852313- [Method of stimulating growth of melanocyte cells by administering stem cell factor](#)
- 7368527- [HADDE71 polypeptides](#)
- 7368531- [Human secreted proteins](#)
- 7399633- [Methods for immortalizing cells](#)
- 7411051- [Antibodies to HDPPA04 polypeptide](#)
- 7415358- [Molecular toxicology modeling](#)
- 7425331- [Protein A methods of use](#)
- 7426441- [Methods for determining renal toxins](#)
- 7447594- [Molecular cardiotoxicology modeling](#)
- 7469185- [Primary rat hepatocyte toxicity modeling](#)
- 7595159- [Prediction of Parkinson's disease using gene expression levels of peripheral blood samples](#)
- 7359748- [Apparatus for total immersion photography](#)
- 7291461- [Methods for identifying small molecules that modulate premature translation termination and nonsense mRNA decay](#)
- 6905827- [Methods and compositions for diagnosing or monitoring auto immune and chronic](#)

[inflammatory diseases](#)

- 6967029- [Method for increasing hematopoietic progenitor cells by stem cell factor](#)
- 6974667- [Gene expression profiles in liver cancer](#)
- 7026121- [Methods and compositions for diagnosing and monitoring transplant rejection](#)
- 7101962- [Antibodies of the P2Y10 receptor useful in altering T lymphocyte function](#)
- 7144731- [SCF antibody compositions and methods of using the same](#)
- 7189507- [Methods of diagnosis of ovarian cancer, compositions and methods of screening for modulators of ovarian cancer](#)
- 7211258- [Protein A compositions and methods of use](#)
- 7226791- [Polynucleotides encoding novel guanylate binding proteins \(GBP's\)](#)
- 7235358- [Methods and compositions for diagnosing and monitoring transplant rejection](#)
- 7598031- [Method for the detection of gene transcripts in blood and uses thereof](#)

\*\*\*

Resources from the [NCBI](#) used in this document, [NCBI's standard disclaimer applies](#).

Nothing in this document should be used in place of personal medical advice from your own qualified medical practitioner. See BioPortfolio.com [User Agreement](#)

Send comments and feedback to:

Peter Barfoot Managing Director, BioPortfolio Ltd.

UK Tel: (+44) 1300 321501

USA Voicemail and Fax: (+1) 415 680 2472

[Peter Barfoot peter.barfoot@bioportfolio.com](mailto:peter.barfoot@bioportfolio.com)

All rights reserved. All other trademarks recognized.

BioPortfolio Limited is registered in England & Wales at Stafford House, 10 Prince of Wales Road, Dorchester, Dorset, DT1 1PW, UK. No.3312883 VAT No. GB 744 6483 10

Copyright 1997-2009 - BioPortfolio Limited.

