

Rothmund-Thomson Syndrome

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Rothmund-Thomson Syndrome is a hereditary benign skin condition originally described by August von Rothmund in 1868; Matthew Sydney Thomson published further descriptions in 1936. There have been several reported cases associated with osteosarcoma, and a hereditary genetic basis, mutations in the DNA Helicase RECQL4, has been implicated in the syndrome. ([From the Wikipedia article Rothmund-Thomson Syndrome](#).)

Recent Publications on Rothmund-Thomson Syndrome:



- [MCM10 mediates RECQ4 association with MCM2-7 helicase complex during DNA replication.](#)
*Mutations in RECQ4, a member of the RecQ family of DNA helicases, have...*6th November, 2009
Department of Therapeutic Radiology, Yale University School of Medicine,- EMBO J. 2009 Oct 7;28(19):3005-14. Epub 2009 Aug 20. ([DOI Direct Link](#))
- [Progeroid syndromes and UV-induced oxidative DNA damage.](#)
*Progeroid syndromes are a group of diseases characterized by signs of...*5th November, 2009
Department of Dermatology, Molecular Oncology and Aging, Eberhard Karls- J Investig Dermatol Symp Proc. 2009 Aug;14(1):8-14. ([DOI Direct Link](#))
- [From the rarest to the most common: insights from progeroid syndromes into skin cancer and aging.](#)
*Despite their rarity, diseases of premature aging, or "progeroid"...*25th September, 2009
The Ronald O. Perelman Department of Dermatology, New York University- J Invest Dermatol. 2009 Oct;129(10):2340-50. Epub 2009 Apr 23. ([DOI Direct Link](#))
- [Direct and indirect roles of RECQL4 in modulating base excision repair capacity.](#)
*RECQL4 is a human RecQ helicase which is mutated in approximately...*2nd July, 2009
Laboratory of Molecular Gerontology, National Institute on Aging, NIH,- Hum Mol Genet. 2009 Sep 15;18(18):3470-83. Epub 2009 Jun 29. ([DOI Direct Link](#))
- [A patient with Baller-Gerold syndrome and midline NK/T lymphoma.](#)
*Three autosomal recessive disorders are associated with mutations in the...*18th June, 2009
Center for Medical Genetics, University Children's Hospital, University- Am J Med Genet A. 2009 Feb 15;149A(4):755-9. ([DOI Direct Link](#))
- [p300-mediated acetylation of the Rothmund-Thomson-syndrome gene product RECQL4 regulates its subcellular localization.](#)
*RECQL4 belongs to the conserved RecQ family of DNA helicases, members of...*12th June, 2009
Department of Biochemistry, Faculty of Medicine, Terrence Donnelly Centre- J Cell Sci. 2009 Apr 15;122(Pt 8):1258-67. Epub 2009 Mar 19. ([DOI Direct Link](#))
- [Recurrent RECQL4 imbalance and increased gene expression levels are associated with structural chromosomal instability in sporadic osteosarcoma.](#)
*Osteosarcoma (OS) is an aggressive bone tumor with complex abnormal...*13th May, 2009

Department of Pathology and Molecular Medicine, Richardson Labs, Queen's- Neoplasia. 2009 Mar;11(3):260-8, 3p following 268.

- [Dual DNA unwinding activities of the Rothmund-Thomson syndrome protein, RECQ4.](#)
*Human RECQ helicases have been linked to distinct clinical diseases with...*28th March, 2009
Department of Therapeutic Radiology, Yale University School of Medicine,- EMBO J. 2009 Mar 4;28(5):568-77. Epub 2009 Jan 29. ([DOI Direct Link](#))
- [Early blistering, poikiloderma, hypohidrosis, alopecia and exocrine pancreatic hypofunction: a peculiar variant of Rothmund-Thomson syndrome?](#)
*A 20-year-old male developed early blistering, poikiloderma, hypohidrosis,...*10th March, 2009
Department of Dermatology, Osaka Medical College, 2-7 Daigaku-cho,- Eur J Dermatol. 2008 Nov-Dec;18(6):632-4. Epub 2008 Oct 27. ([DOI Direct Link](#))
- [RecQ family helicases in genome stability: lessons from gene disruption studies in DT40 cells.](#)
*Cells of all living organisms have evolved complex mechanisms to maintain...*24th February, 2009
Molecular Cell Biology Laboratory, Graduate School of Pharmaceutical- Cell Cycle. 2008 Aug 15;7(16):2472-8. Epub 2008 Aug 18.
- [The mutation spectrum in RECQL4 diseases.](#)
*Mutations in the RECQL4 gene can lead to three clinical phenotypes with...*21st February, 2009
Department of Molecular Medicine, National Public Health Institute and- Eur J Hum Genet. 2009 Feb;17(2):151-8. Epub 2008 Aug 20. ([DOI Direct Link](#))
- [Rising from the RecQ-age: the role of human RecQ helicases in genome maintenance.](#)
*The RecQ helicases are guardians of the genome. Members of this conserved...*27th January, 2009
Laboratory of Molecular Gerontology, National Institute on Aging, National- Trends Biochem Sci. 2008 Dec;33(12):609-20. Epub 2008 Oct 14. ([DOI Direct Link](#))
- [Poikiloderma with neutropenia, Clericuzio type, in a family from Morocco.](#)
*Three siblings from Morocco consanguineous family presented with cutaneous...*17th December, 2008
Department of Pediatric Dermatology (National Reference Center for Rare- Am J Med Genet A. 2008 Nov 1;146A(21):2762-9. ([DOI Direct Link](#))
- [Drosophila homologue of the Rothmund-Thomson syndrome gene: essential function in DNA replication during development.](#)
*Members of the RecQ family play critical roles in maintaining genome...*17th December, 2008
Department of Biochemistry, Duke University Medical Center, Durham, NC- Dev Biol. 2008 Nov 1;323(1):130-42. Epub 2008 Aug 15. ([DOI Direct Link](#))
- [RecQ4 facilitates UV light-induced DNA damage repair through interaction with nucleotide excision repair factor xeroderma pigmentosum group A \(XPA\).](#)
*Mutations in the RECQL4 helicase gene have been linked to Rothmund-Thomson...*17th December, 2008
Department of Cancer Biology and the Cancer Center, University of- J Biol Chem. 2008 Oct 24;283(43):29037-44. Epub 2008 Aug 7. ([DOI Direct Link](#))

BioNews Results for Rothmund-Thomson Syndrome

- [UPDATE 2-Sequenom posts wider Q3 net loss; shares down](#)
Forbes.com: Nov 10 2009 12:07AM Matching: thomson syndrome
- [UPDATE 2-Sequenom posts wider Q3 net loss; shares down](#)
Forbes.com: Nov 10 2009 12:07AM Matching: thomson syndrome
- [UPDATE 2-Sequenom posts wider Q3 net loss; shares down](#)
Interactive Investor International: Nov 9 2009 11:53PM Matching: thomson syndrome



Rothmund-Thomson Syndrome Patents:

- 7608392- [Diagnostic assay for measuring a cell mediated immune response](#)
- 6436706- [Antisense inhibitor of RECQL4 expression](#)
- 6977162- [Rapid analysis of variations in a genome](#)
- 7081522- [Gene causative of Rothmund-Thomson syndrome and gene product](#)
- 7172865- [Rapid and sensitive assay for the detection and quantification of coregulators of nucleic acid binding factors](#)
- 7208274- [Rapid analysis of variations in a genome](#)
- 7329497- [Gene causative of Rothmund-Thomson syndrome and its gene product](#)
- 7332277- [Methods for detection of genetic disorders](#)
- 7359748- [Apparatus for total immersion photography](#)
- 7442506- [Methods for detection of genetic disorders](#)
- 7541511- [Mouse exhibiting characteristics of Rothmund-Thomson syndrome and preparation method thereof](#)
- 7576192- [Rapid and sensitive assay for the detection and quantification of coregulators of nucleic acid binding factors](#)
- 7598060- [Rapid analysis of variations in a genome](#)
- 6387633- [Method for identifying unstable genes](#)

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