Inderjeet Dokal

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10267023/publications.pdf

Version: 2024-02-01

112 papers	11,598 citations	30070 54 h-index	29157 104 g-index
112 all docs	112 docs citations	112 times ranked	8927 citing authors

#	Article	IF	Citations
1	High-throughput STELA provides a rapid test for the diagnosis of telomere biology disorders. Human Genetics, 2021, 140, 945-955.	3.8	12
2	Genome-wide whole-blood transcriptome profiling across inherited bone marrow failure subtypes. Blood Advances, 2021, 5, 5360-5371.	5.2	1
3	Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. Blood, 2021, 138, 864-864.	1.4	2
4	A frameshift variant in specificity protein 1 triggers superactivation of Sp1-mediated transcription in familial bone marrow failure. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 17151-17155.	7.1	2
5	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	12.8	81
6	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the ClinicalÂDiagnosis of Inborn Errors of Immunity. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1763-1770.	3.8	381
7	Germline NPM1 mutations lead to altered rRNA 2â€²-O-methylation and cause dyskeratosis congenita. Nature Genetics, 2019, 51, 1518-1529.	21.4	84
8	Expanding the phenotypic and genetic spectrum of radioulnar synostosis associated hematological disease. Haematologica, 2018, 103, e284-e287.	3.5	17
9	British Committee for Standards in Haematology guidelines for aplastic anemia: Single centre retrospective review finds no compelling evidence for the recommended higher platelet count threshold of 20ÂÃ—Â10⁹/L â€•<scp>RESPONSE</scp> to Yan etÂal. British Journal of Haematology, 2018, 182, 286-287.	2.5	1
10	Genome instability is a consequence of transcription deficiency in patients with bone marrow failure harboring biallelic ERCC6L2 variants. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 7777-7782.	7.1	37
11	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. Leukemia, 2018, 32, 2502-2507.	7.2	48
12	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenitaâ€“like phenotypes. Blood, 2018, 132, 1349-1353.	1.4	16
13	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	3.5	15
14	TGF-Î² Pathway Inhibition Signals New Hope for Fanconi Anemia. Cell Stem Cell, 2016, 18, 567-568.	11.1	5
15	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. Haematologica, 2016, 101, 1180-1189.	3.5	34
16	Guidelines for the diagnosis and management of adult aplastic anaemia. British Journal of Haematology, 2016, 172, 187-207.	2.5	539
17	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. American Journal of Human Genetics, 2016, 99, 115-124.	6.2	85
18	Disease evolution and outcomes in familial AML with germline CEBPA mutations. Blood, 2015, 126, 1214-1223.	1.4	157

#	Article	IF	Citations
19	Triallelic and epigenetic-like inheritance in human disorders of telomerase. Blood, 2015, 126, 176-184.	1.4	49
20	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Journal of Clinical Investigation, 2015, 125, 2151-2160.	8.2	165
21	Inherited bone marrow failure syndromes. Hematology, 2015, 20, 433-434.	1.5	13
22	Clinical utility gene card for: Dyskeratosis congenita â€“ update 2015. European Journal of Human Genetics, 2015, 23, 558-558.	2.8	63
23	Dyskeratosis Congenita. , 2014, , 267-280.		1
24	ERCC6L2 Mutations Link a Distinct Bone-Marrow-Failure Syndrome to DNA Repair and Mitochondrial Function. American Journal of Human Genetics, 2014, 94, 246-256.	6.2	58
25	Overlap of Syndromes Associated with Myelodysplasia and Leukemia. Blood, 2014, 124, SCI-32-SCI-32.	1.4	0
26	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. American Journal of Human Genetics, 2013, 92, 448-453.	6.2	191
27	Haematological recovery in dyskeratosis congenita patients treated with danazol. British Journal of Haematology, 2013, 162, 854-856.	2.5	49
28	Aberrant 3â€² oligoadenylation of spliceosomal U6 small nuclear RNA in poikiloderma with neutropenia. Blood, 2013, 121, 1028-1038.	1.4	65
29	Heightened DNA damage response impairs hematopoiesis in Fanconi anemia. Haematologica, 2012, 97, 1117-1117.	3.5	5
30	Exome sequencing identifies MPL as a causative gene in familial aplastic anemia. Haematologica, 2012, 97, 524-528.	3.5	42
31	Limbal stem cell deficiency in patients with inherited stem cell disorder of dyskeratosis congenita. International Ophthalmology, 2012, 32, 615-622.	1.4	16
32	Revertant Somatic Mosaicism by Mitotic Recombination in Dyskeratosis Congenita. American Journal of Human Genetics, 2012, 90, 426-433.	6.2	97
33	Exome Sequencing Identifies Autosomal-Dominant SRP72 Mutations Associated with Familial Aplasia and Myelodysplasia. American Journal of Human Genetics, 2012, 90, 888-892.	6.2	94
34	Marked genetic heterogeneity in familial myelodysplasia/acute myeloid leukaemia. British Journal of Haematology, 2012, 158, 242-248.	2.5	65
35	Prenatal and postnatal growth retardation, microcephaly, developmental delay, and pigmentation abnormalities: Naegeli syndrome, dyskeratosis congenita, poikiloderma Clericuzio type, or separate entity?. European Journal of Medical Genetics, 2011, 54, 231-235.	1.3	6
36	Differences in Disease Severity but Similar Telomere Lengths in Genetic Subgroups of Patients with Telomerase and Shelterin Mutations. PLoS ONE, 2011, 6, e24383.	2.5	79

#	Article	IF	Citations
37	Dyskeratosis congenita and the DNA damage response. British Journal of Haematology, 2011, 153, 634-643.	2.5	29
38	Clinical utility gene card for: Dyskeratosis congenita. European Journal of Human Genetics, 2011, 19, 3-4.	2.8	24
39	Inherited bone marrow failure syndromes. Journal of Hematopathology, 2011, 4, 53-60.	0.4	5
40	Dyskeratosis Congenita. Hematology American Society of Hematology Education Program, 2011, 2011, 480-486.	2.5	236
41	Inherited bone marrow failure syndromes. Haematologica, 2010, 95, 1236-1240.	3.5	105
42	Emberger syndromeâ€"Primary lymphedema with myelodysplasia: Report of seven new cases. American Journal of Medical Genetics, Part A, 2010, 152A, 2287-2296.	1.2	70
43	Mutations in C16orf57 and normal-length telomeres unify a subset of patients with dyskeratosis congenita, poikiloderma with neutropenia and Rothmundâ€"Thomson syndrome. Human Molecular Genetics, 2010, 19, 4453-4461.	2.9	87
44	Severe Variant of Xâ€linked Dyskeratosis Congenita (HoyeraalâHreidarsson Syndrome) Causes Significant Enterocolitis in Early Infancy. Journal of Pediatric Gastroenterology and Nutrition, 2009, 49, 359-363.	1.8	23
45	Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia. Human Mutation, 2009, 30, 1567-1573.	2.5	107
46	Dyskeratosis congenita: The first NIH clinical research workshop. Pediatric Blood and Cancer, 2009, 53, 520-523.	1.5	66
47	Exogenous TERC alone can enhance proliferative potential, telomerase activity and telomere length in lymphocytes from dyskeratosis congenita patients. British Journal of Haematology, 2009, 144, 771-781.	2.5	31
48	Advances in the understanding of dyskeratosis congenita. British Journal of Haematology, 2009, 145, 164-172.	2.5	141
49	Guidelines for the diagnosis and management of aplastic anaemia. British Journal of Haematology, 2009, 147, 43-70.	2.5	487
50	Single-Molecule Analysis of the Human Telomerase RNAÂ·Dyskerin Interaction and the Effect of Dyskeratosis Congenita Mutations. Biochemistry, 2009, 48, 10858-10865.	2.5	21
51	Dyskeratosis congenita, stem cells and telomeres. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 371-379.	3.8	139
52	Understanding aplastic anaemia/bone-marrow failure syndromes. Paediatrics and Child Health (United) Tj ETQq0 0 0 rgBT /Overlock 10 T	0.4	5
53	Hepatitis-associated Aplastic Anemia Presenting as a Familial Bone Marrow Failure Syndrome. Journal of Pediatric Hematology/Oncology, 2009, 31, 884-887.	0.6	7
54	Ataxia and pancytopenia caused by a mutation in TINF2. Human Genetics, 2008, 124, 507-513.	3.8	26

#	Article	IF	Citations
55	Inherited aplastic anaemias/bone marrow failure syndromes. Blood Reviews, 2008, 22, 141-153.	5.7	92
56	Circulating haematopoietic progenitors are differentially reduced amongst subtypes of dyskeratosis congenita. British Journal of Haematology, 2008, 140, 719-722.	2.5	8
57	Dyskeratosis Congenita: A historical perspective. Mechanisms of Ageing and Development, 2008, 129, 48-59.	4.6	65
58	Mutations in the telomerase component NHP2 cause the premature ageing syndrome dyskeratosis congenita. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 8073-8078.	7.1	294
59	Dissecting â€œstressâ€•in Fanconi anemia. Blood, 2008, 111, 1756-1757.	1.4	0
60	TINF2 mutations result in very short telomeres: analysis of a large cohort of patients with dyskeratosis congenita and related bone marrow failure syndromes. Blood, 2008, 112, 3594-3600.	1.4	284
61	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica, 2008, 93, 943-944.	3.5	43
62	Low Dose Alemtuzumab Achieves Long-Term Engraftment with Low Level Mixed Chimerism in Related Haemopoietic Stem Cell Transplantation for Haemoglobinopathies. Blood, 2008, 112, 3304-3304.	1.4	0
63	Full Donor Engraftment without Chronic GvHD Following HLA-Matched Sibling Donor Haemopoietic Stem Cell Transplantation for Diamond Blackfan Anaemia. Blood, 2008, 112, 4394-4394.	1.4	0
64	Fanconi anemia is a highly penetrant cancer susceptibility syndrome. Haematologica, 2008, 93, 486-8.	3.5	7
65	Genetic heterogeneity in autosomal recessive dyskeratosis congenita with one subtype due to mutations in the telomerase-associated protein NOP10. Human Molecular Genetics, 2007, 16, 1619-1629.	2.9	285
66	Functional characterization of novel telomerase RNA (TERC) mutations in patients with diverse clinical and pathological presentations. Haematologica, 2007, 92, 1013-1020.	3.5	58
67	Telomerase reverse-transcriptase homozygous mutations in autosomal recessive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. Blood, 2007, 110, 4198-4205.	1.4	155
68	The Loss of Telomerase Activity in Highly Differentiated CD8+CD28â^'CD27â^' T Cells Is Associated with Decreased Akt (Ser473) Phosphorylation. Journal of Immunology, 2007, 178, 7710-7719.	0.8	185
69	Dyskeratosis congenita: Advances in the understanding of the telomerase defect and the role of stem cell transplantation. Pediatric Transplantation, 2007, 11, 584-594.	1.0	109
70	Dyskeratosis Congenita. Seminars in Hematology, 2006, 43, 157-166.	3.4	116
71	Diagnostic radioisotopes in haematology. , 2006, , 357-378.		0
72	Mutations in dyskeratosis congenita: their impact on telomere length and the diversity of clinical presentation. Blood, 2006, 107, 2680-2685.	1.4	310

#	Article	IF	Citations
73	Fanconi's anaemia and related bone marrow failure syndromes. British Medical Bulletin, 2006, 77-78, 37-53.	6.9	31
74	Dyskeratosis congenita: a disorder of telomerase deficiency and its relationship to other diseases. Expert Review of Dermatology, 2006, 1, 463-479.	0.3	7
75	Dyskeratosis Congenita: A Disorder of Defective Telomere Maintenance?. International Journal of Hematology, 2005, 82, 184-189.	1.6	40
76	A human ortholog of archaeal DNA repair protein Hef is defective in Fanconi anemia complementation group M. Nature Genetics, 2005, 37, 958-963.	21.4	395
77	Peripheral neuropathyâ€"a novel finding in dyskeratosis congenita. European Journal of Paediatric Neurology, 2005, 9, 85-89.	1.6	8
78	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. Blood Cells, Molecules, and Diseases, 2005, 34, 257-263.	1.4	174
79	Dyskeratosis congenita: telomerase, telomeres and anticipation. Current Opinion in Genetics and Development, 2005, 15, 249-257.	3.3	113
80	Dyskeratosis congenita: molecular insights into telomerase function, ageing and cancer. Expert Reviews in Molecular Medicine, 2004, 6, 1-23.	3.9	41
81	Fanconi anaemia and leukaemia â€" clinical and molecular aspects. British Journal of Haematology, 2004, 126, 176-191.	2.5	124
82	Disease anticipation is associated with progressive telomere shortening in families with dyskeratosis congenita due to mutations in TERC. Nature Genetics, 2004, 36, 447-449.	21.4	425
83	Total Deletion of in Vivo Telomere Elongation Capacity: An Ambitious but Possibly Ultimate Cure for All Ageâ€Related Human Cancers. Annals of the New York Academy of Sciences, 2004, 1019, 147-170.	3.8	20
84	Telomerase dysfunction and dyskeratosis congenita. Cytotechnology, 2004, 45, 13-22.	1.6	3
85	A mutation in a functional Sp1 binding site of the telomerase RNA gene (hTERC) promoter in a patient with Paroxysmal Nocturnal Haemoglobinuria. BMC Hematology, 2004, 4, 3.	2.6	28
86	Heterozygous telomerase RNA mutations found in dyskeratosis congenita and aplastic anemia reduce telomerase activity via haploinsufficiency. Blood, 2004, 104, 3936-3942.	1.4	122
87	Further delineation of the congenital form of X-linked dyskeratosis congenita (Hoyeraal-Hreidarsson) Tj ETQq1 1 0.784314 rgBT /Over	2.7	70
88	Two brothers with findings resembling congenital intrauterine infection-like syndrome (pseudo-TORCH syndrome). American Journal of Medical Genetics Part A, 2003, 120A, 261-265.	2.4	32
89	Dyskeratosis congenita: its link to telomerase and aplastic anaemia. Blood Reviews, 2003, 17, 217-225.	5.7	90
90	Bone marrow transplantation for Î²-thalassaemia major: the UK experience in two paediatric centres. British Journal of Haematology, 2003, 120, 289-295.	2.5	85

#	Article	IF	Citations
91	Inherited aplastic anaemia. The Hematology Journal, 2003, 4, 3-9.	1.4	17
92	Association between aplastic anaemia and mutations in telomerase RNA. Lancet, The, 2002, 359, 2168-2170.	13.7	261
93	Detection of implanted splenic tissue using 99m-Technetium-labelled heat-damaged autologous red cells. British Journal of Haematology, 2002, 118, 2-2.	2.5	0
94	A novel DKC1 mutation, severe combined immunodeficiency (T+ B- NK- SCID) and bone marrow transplantation in an infant with Hoyeraal-Hreidarsson syndrome. British Journal of Haematology, 2002, 119, 765-768.	2.5	69
95	Targeted disruption of Dkc1, the gene mutated in X-linked dyskeratosis congenita, causes embryonic lethality in mice. Oncogene, 2002, 21, 7740-7744.	5.9	93
96	Very Short Telomeres in the Peripheral Blood of Patients with X-Linked and Autosomal Dyskeratosis Congenita. Blood Cells, Molecules, and Diseases, 2001, 27, 353-357.	1.4	196
97	A disease of premature ageing. Lancet, The, 2001, 358, S27.	13.7	42
98	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. Human Genetics, 2001, 108, 299-303.	3.8	77
99	The RNA component of telomerase is mutated in autosomal dominant dyskeratosis congenita. Nature, 2001, 413, 432-435.	27.8	902
100	Dyskeratosis congenita in all its forms. British Journal of Haematology, 2000, 110, 768-779.	2.5	487
101	The Inherited Bone Marrow Failure Syndromes: Fanconi Anemia, Dyskeratosis Congenita and Diamondâ€Blackfan Anemia. Reviews in Clinical and Experimental Hematology, 2000, 4, 183-215.	0.1	4
102	The genetics of Fanconi's anaemia. Best Practice and Research in Clinical Haematology, 2000, 13, 407-425.	1.7	31
103	Engraftment of Hematopoietic Progenitor Cells Transduced with the Fanconi Anemia Group C Gene (FANCC). Human Gene Therapy, 1999, 10, 2337-2346.	2.7	127
104	X-linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolar functions. Nature Genetics, 1998, 19, 32-38.	21.4	867
105	Chromosomal breakage analysis in dyskeratosis congenita peripheral blood lymphocytes. British Journal of Haematology, 1998, 102, 1162-1164.	2.5	33
106	Dyskeratosis Congenita (DC) Registry: identification of new features of DC. British Journal of Haematology, 1998, 103, 990-996.	2.5	165
107	Adult onset of acute myeloid leukaemia (M6) in patients with Shwachmanâ€Diamond syndrome. British Journal of Haematology, 1997, 99, 171-173.	2.5	59
108	Dyskeratosis congenita: an inherited bone marrow failure syndrome. British Journal of Haematology, 1996, 92, 775-779.	2.5	67

#	Article	IF	Citations
109	Severe aplastic anemia including FanconiÊ¼s anemia and dyskeratosis congenita. Current Opinion in Hematology, 1996, 3, 453-460.	2.5	13
110	Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutation, 1996, 8, 140-148.	2.5	42
111	Dyskeratosis Congenita Is a Chromosomal Instability Disorder. Leukemia and Lymphoma, 1994, 15, 1-7.	1.3	40
112	Detection of hereditary haemochromatosis in an HLA-identical pedigree showing discordance between HLA class I genes and the disease locus. Human Genetics, 1991, 88, 209-14.	3.8	3