Tom Vulliamy

List of Publications by Year
in descending order

52 papers	5,584 citations	147801 31 h-index	197818 49 g-index
52 all docs	52 docs citations	52 times ranked	6552 citing authors

#	Article	IF	Citations
1	The RNA component of telomerase is mutated in autosomal dominant dyskeratosis congenita. Nature, 2001, 413, 432-435.	27.8	902
2	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
3	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
4	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
5	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
6	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
7	Association between aplastic anaemia and mutations in telomerase RNA. Lancet, The, 2002, 359, 2168-2170.	13.7	261
8	Isolation of human glucose-6-pbosphate debydrogenase (G6PD) cDNA clones: primary structure of the protein and unusual 5' non-coding region. Nucleic Acids Research, 1986, 14, 2511-2522.	14.5	242
9	Telomerase reverse-transcriptase homozygous mutations in autosomal recessive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. Blood, 2007, 110, 4198-4205.	1.4	155
10	Heterozygous telomerase RNA mutations found in dyskeratosis congenita and aplastic anemia reduce telomerase activity via haploinsufficiency. Blood, 2004, 104, 3936-3942.	1.4	122
11	Dyskeratosis Congenita. Seminars in Hematology, 2006, 43, 157-166.	3.4	116
12	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. Blood Advances, 2019, 3, 2962-2979.	5.2	110
13	Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia. Human Mutation, 2009, 30, 1567-1573.	2.5	107
14	Inherited bone marrow failure syndromes. Haematologica, 2010, 95, 1236-1240.	3.5	105
15	Revertant Somatic Mosaicism by Mitotic Recombination in Dyskeratosis Congenita. American Journal of Human Genetics, 2012, 90, 426-433.	6.2	97
16	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
17	Inherited aplastic anaemias/bone marrow failure syndromes. Blood Reviews, 2008, 22, 141-153.	5.7	92
18	Dyskeratosis congenita: its link to telomerase and aplastic anaemia. Blood Reviews, 2003, 17, 217-225.	5.7	90

#	Article	IF	Citations
19	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
20	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
21	Variants of glucose-6-phosphate dehydrogenase are due to missense mutations spread throughout the coding region of the gene. Human Mutation, 1993, 2, 159-167.	2.5	83
22	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. Human Genetics, 2001, 108, 299-303.	3.8	77
23	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
24	Marked genetic heterogeneity in familial myelodysplasia/acute myeloid leukaemia. British Journal of Haematology, 2012, 158, 242-248.	2.5	65
25	Clinical utility gene card for: Dyskeratosis congenita â€“ update 2015. European Journal of Human Genetics, 2015, 23, 558-558.	2.8	63
26	Functional characterization of novel telomerase RNA (TERC) mutations in patients with diverse clinical and pathological presentations. Haematologica, 2007, 92, 1013-1020.	3.5	58
27	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
28	Air pollution, ethnicity and telomere length in east London schoolchildren: An observational study. Environment International, 2016, 96, 41-47.	10.0	44
29	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica, 2008, 93, 943-944.	3.5	43
30	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
31	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
32	Deficiency in red blood cells. Nature, 1991, 350, 115-115.	27.8	31
33	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
34	Dyskeratosis congenita and the DNA damage response. British Journal of Haematology, 2011, 153, 634-643.	2.5	29
35	Laboratory diagnosis of G6PD deficiency. A British Society for Haematology Guideline. British Journal of Haematology, 2020, 189, 24-38.	2.5	29
36	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

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37	Relative telomere lengths in tumor and normal mucosa are related to disease progression and chromosome instability profiles in colorectal cancer. Oncotarget, 2016, 7, 36474-36488.	1.8	23
38	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
39	Independent origin of single and double mutations in the human glucose 6-phosphate dehydrogenase gene. Human Mutation, 1996, 8, 311-318.	2.5	18
40	Expanding the phenotypic and genetic spectrum of radioulnar synostosis associated hematological disease. Haematologica, 2018, 103, e284-e287.	3.5	17
41	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenitaâ€“like phenotypes. Blood, 2018, 132, 1349-1353.	1.4	16
42	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	3.5	15
43	High-throughput STELA provides a rapid test for the diagnosis of telomere biology disorders. Human Genetics, 2021, 140, 945-955.	3.8	12
44	DNA Genotypic Conservation During Phenotypic Switch from T-cell Acute Lymphoblastic Leukaemia to Acute Myeloblastic Leukaemia. Leukemia and Lymphoma, 1989, 1, 21-28.	1.3	11
45	Characterization of G6PD deficiency in southern Croatia: description of a new variant, G6PD Split. Journal of Human Genetics, 2005, 50, 547-549.	2.3	11
46	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
47	GENETIC VARIATION OF HUMAN ERYTHROCYTE GLUCOSE-6-PHOSPHATE DEHYDROGENASE. , 2000, , 251-275.		2
48	Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. Blood, 2021, 138, 864-864.	1.4	2
49	Nonâ€lymphoid blast crisis of CML with rearrangement of immunoglobulin and Tâ€cell receptor delta genes. European Journal of Haematology, 1991, 47, 36-41.	2.2	1
50	Genome-wide whole-blood transcriptome profiling across inherited bone marrow failure subtypes. Blood Advances, 2021, 5, 5360-5371.	5.2	1
51	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
52	Telomere Lengths Correlate With Inflammatory Markers In Sickle Cell Disease. Blood, 2013, 122, 2230-2230.	1.4	0