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	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	Laboratory diagnosis of G6PD deficiency. A British Society for Haematology Guideline. British Journal of Haematology, 2020, 189, 24-38.	2.5	29
6	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. Blood Advances, 2019, 3, 2962-2979.	5.2	110
7	Expanding the phenotypic and genetic spectrum of radioulnar synostosis associated hematological disease. Haematologica, 2018, 103, e284-e287.	3.5	17
8	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenitaâ€“like phenotypes. Blood, 2018, 132, 1349-1353.	1.4	16
9	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
10	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
11	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	3.5	15
12	Air pollution, ethnicity and telomere length in east London schoolchildren: An observational study. Environment International, 2016, 96, 41-47.	10.0	44
13	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
14	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
15	Clinical utility gene card for: Dyskeratosis congenita â€“ update 2015. European Journal of Human Genetics, 2015, 23, 558-558.	2.8	63
16	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
17	Telomere Lengths Correlate With Inflammatory Markers In Sickle Cell Disease. Blood, 2013, 122, 2230-2230.	1.4	0
18	Revertant Somatic Mosaicism by Mitotic Recombination in Dyskeratosis Congenita. American Journal of Human Genetics, 2012, 90, 426-433.	6.2	97

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19	Marked genetic heterogeneity in familial myelodysplasia/acute myeloid leukaemia. British Journal of Haematology, 2012, 158, 242-248.	2.5	65
20	Dyskeratosis congenita and the DNA damage response. British Journal of Haematology, 2011, 153, 634-643.	2.5	29
21	Inherited bone marrow failure syndromes. Haematologica, 2010, 95, 1236-1240.	3.5	105
22	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
23	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
24	Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia. Human Mutation, 2009, 30, 1567-1573.	2.5	107
25	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
26	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
27	Inherited aplastic anaemias/bone marrow failure syndromes. Blood Reviews, 2008, 22, 141-153.	5.7	92
28	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
29	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
30	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica, 2008, 93, 943-944.	3.5	43
31	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
32	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
33	Functional characterization of novel telomerase RNA (TERC) mutations in patients with diverse clinical and pathological presentations. Haematologica, 2007, 92, 1013-1020.	3.5	58
34	Telomerase reverse-transcriptase homozygous mutations in autosomal recessive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. Blood, 2007, 110, 4198-4205.	1.4	155
35	Dyskeratosis Congenita. Seminars in Hematology, 2006, 43, 157-166.	3.4	116
36	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

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37	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
38	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
39	Heterozygous telomerase RNA mutations found in dyskeratosis congenita and aplastic anemia reduce telomerase activity via haploinsufficiency. Blood, 2004, 104, 3936-3942.	1.4	122
40	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
41	Dyskeratosis congenita: its link to telomerase and aplastic anaemia. Blood Reviews, 2003, 17, 217-225.	5.7	90
42	Association between aplastic anaemia and mutations in telomerase RNA. Lancet, The, 2002, 359, 2168-2170.	13.7	261
43	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
44	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. Human Genetics, 2001, 108, 299-303.	3.8	77
45	The RNA component of telomerase is mutated in autosomal dominant dyskeratosis congenita. Nature, 2001, 413, 432-435.	27.8	902
46	GENETIC VARIATION OF HUMAN ERYTHROCYTE GLUCOSE-6-PHOSPHATE DEHYDROGENASE. , 2000, , 251-275.		2
47	Independent origin of single and double mutations in the human glucose 6-phosphate dehydrogenase gene. Human Mutation, 1996, 8, 311-318.	2.5	18
48	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
49	Deficiency in red blood cells. Nature, 1991, 350, 115-115.	27.8	31
50	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
51	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
52	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