Raymond Dalgleish

List of Publications by Year
in descending order

73 papers	3,954 citations	218677 26 h-index	123424 61 g-index
77 all docs	77 docs citations	77 times ranked	6689 citing authors

#	Article	IF	Citations
1	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	2.5	1,194
2	Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Human Mutation, 2007, 28, 209-221.	2.5	620
3	The human type I collagen mutation database. Nucleic Acids Research, 1997, 25, 181-187.	14.5	282
4	The Human Collagen Mutation Database 1998. Nucleic Acids Research, 1998, 26, 253-255.	14.5	198
5	Human type III collagen gene expression is coordinately modulated with the type I collagen genes during fibroblast growth. Biochemistry, 1986, 25, 1408-1413.	2.5	119
6	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
7	VariantValidator: Accurate validation, mapping, and formatting of sequence variation descriptions. Human Mutation, 2018, 39, 61-68.	2.5	105
8	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	8.2	100
9	Chromosomal assignments of the genes coding for human types II, III, and IV collagen: a dispersed gene family.. Proceedings of the National Academy of Sciences of the United States of America, 1985, 82, 3330-3334.	7.1	82
10	Isolation and partial sequence of recombinant plasmids containing human Î±-, Î²- and Î³-globin cDNA fragments. Nature, 1978, 273, 640-643.	27.8	78
11	Length polymorphism in the threonine-glycine-encoding repeat region of theperiod gene inDrosophila. Journal of Molecular Evolution, 1991, 32, 238-246.	1.8	74
12	Locus Reference Genomic: reference sequences for the reporting of clinically relevant sequence variants. Nucleic Acids Research, 2014, 42, D873-D878.	14.5	73
13	Clinical utility gene card for: osteogenesis imperfecta. European Journal of Human Genetics, 2013, 21, 1-4.	2.8	68
14	Linkage of a polymorphic marker for the type III collagen gene (COL3A1) to atypical autosomal dominant Ehlers-Danlos syndrome type IV in a large Belgian pedigree. Human Genetics, 1988, 78, 276-281.	3.8	50
15	Novel variants in the SOHLH2 gene are implicated in human premature ovarian failure. Fertility and Sterility, 2014, 101, 1104-1109.e6.	1.0	50
16	Guidelines for establishing locus specific databases. Human Mutation, 2012, 33, 298-305.	2.5	48
17	HUGO Gene Nomenclature Committee (HGNC) recommendations for the designation of gene fusions. Leukemia, 2021, 35, 3040-3043.	7.2	42
18	Curating gene variant databases (LSDBs): Toward a universal standard. Human Mutation, 2012, 33, 291-297.	2.5	41

#	Article	IF	Citations
19	Quantifying the use of bioresources for promoting their sharing in scientific research. GigaScience, 2013, 2, 7.	6.4	38
20	Genetic counselling on brittle grounds: Recurring osteogenesis imperfecta due to parental mosaicism for a dominant mutation. European Journal of Pediatrics, 1995, 154, 123-129.	2.7	37
21	Practical guidelines addressing ethical issues pertaining to the curation of human locus-specific variation databases (LSDBs). Human Mutation, 2010, 31, 1179-1184.	2.5	36
22	EMQN best practice guidelines for genetic testing in dystrophinopathies. European Journal of Human Genetics, 2020, 28, 1141-1159.	2.8	35
23	An RFLP associated with the human type III collagen gene (COL3A1). Nucleic Acids Research, 1985, 13, 4609-4609.	14.5	28
24	Transcription factor SOHLH1 potentially associated with primary ovarian insufficiency. Fertility and Sterility, 2015, 103, 548-553.e5.	1.0	28
25	Substitution of glycine-661 by serine in the Î±1(I) and Î±2(1) chains of type I collagen results in different clinical and biochemical phenotypes. Human Genetics, 1996, 97, 324-329.	3.8	26
26	The collÎ±gen III fibril has a â€œflexi-rodâ€•structure of flexible sequences interspersed with rigid bioactive domains including two with hemostatic roles. PLoS ONE, 2017, 12, e0175582.	2.5	24
27	Length polymorphism in the pro ?2(I) collagen gene: an alternative explanation in a case of Marfan syndrome. Human Genetics, 1986, 73, 91-92.	3.8	23
28	Cafe Variome: General-Purpose Software for Making Genotype-Phenotype Data Discoverable in Restricted or Open Access Contexts. Human Mutation, 2015, 36, 957-964.	2.5	23
29	WAVe: web analysis of the variome. Human Mutation, 2011, 32, 729-734.	2.5	20
30	hgvs: A Python package for manipulating sequence variants using HGVS nomenclature: 2018 Update. Human Mutation, 2018, 39, 1803-1813.	2.5	20
31	Characterization of the human secreted phosphoprotein 24 gene (SPP2) and comparison of the protein sequence in nine species. Matrix Biology, 2004, 22, 641-651.	3.6	19
32	VarioML framework for comprehensive variation data representation and exchange. BMC Bioinformatics, 2012, 13, 254.	2.6	17
33	Dissecting the phenotypic variability of osteogenesis imperfecta. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	16
34	A single amino acid deletion in the ?2(I) chain of type I collagen produces osteogenesis imperfecta type III. Human Genetics, 1993, 90, 621-8.	3.8	15
35	A Gly859Ser substitution in the triple helical domain of the Î±2 chain of type I collagen resulting in osteogenesis imperfecta type III in two unrelated individuals. Human Mutation, 1994, 3, 391-394.	2.5	14
36	Human pro Î± 1(III) collagen: cDNA sequence for the 3â€² end. Nucleic Acids Research, 1988, 16, 2337-2337.	14.5	13

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37	SSCP detection of a Gly565Val substitution in the pro?(I) collagen chain resulting in osteogenesis imperfecta type II. Human Genetics, 1993, 91, 439-44.	3.8	13
38	Ethnic specificity of variants of the ESR1, HK3, BRSK1 genes and the 8q22.3 locus: No association with premature ovarian failure (POF) in Serbian women. Maturitas, 2014, 77, 64-67.	2.4	12
39	A Gly238Ser substitution in the ?2 chain of type I collagen results in osteogenesis imperfecta type III. Human Genetics, 1995, 95, 215-8.	3.8	10
40	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
41	Three unrelated individuals with perinatally lethal osteogenesis imperfecta resulting from identical Gly502Ser substitutions in the Î±2-chain of type I collagen. Human Genetics, 1994, 94, 497-503.	3.8	9
42	Lack of association betweenESR1gene polymorphisms and premature ovarian failure in Serbian women. Climacteric, 2014, 17, 247-251.	2.4	9
43	Substitution of glycine-172 by arginine in the Î±1 chain of type I collagen in a patient with osteogenesis imperfecta, type III. Human Mutation, 1994, 3, 324-326.	2.5	8
44	Ehlersâ€Danlos syndrome type IV caused by Gly400Glu, Gly595Cys and Glyl003Asp substitutions in collagen III: clinical features, biochemical screening, and molecular confirmation. Clinical Genetics, 1996, 49, 286-295.	2.0	7
45	Variobox: Automatic Detection and Annotation of Human Genetic Variants. Human Mutation, 2014, 35, 202-207.	2.5	7
46	LSDBs and How They Have Evolved. Human Mutation, 2016, 37, 532-539.	2.5	6
47	PCR detection of a 38 bp length variant in the COL1A2 gene. Nucleic Acids Research, 1990, 18, 5925-5925.	14.5	5
48	A common classification framework for histone sequence alterations in tumours: an expert consensus proposal. Journal of Pathology, 2021, 254, 109-120.	4.5	5
49	The divergence between human and baboon globin genes. Nucleic Acids and Protein Synthesis, 1976, 435, 76-81.	1.7	4
50	PCR detection of a COL1A1RsalRFLP. Nucleic Acids Research, 1991, 19, 3163-3163.	14.5	4
51	AHaeIII RFLP in COL1A1. Nucleic Acids Research, 1990, 18, 5926-5926.	14.5	3
52	Collagen gene structure. Biochemical Society Transactions, 1988, 16, 661-663.	3.4	2
53	Bilateral consecutive rupture of the quadriceps tendon in a man with BstUI polymorphism of the COL5A1 gene. Knee Surgery, Sports Traumatology, Arthroscopy, 2011, 19, 1403-1403.	4.2	2
54	The Human Variome Project: ensuring the quality of DNA variant databases in inherited renal disease. Pediatric Nephrology, 2015, 30, 1893-1901.	1.7	2

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55	Substitution of glycine-661 by serine in the Î±1(I) and Î±2(I) chains of type I collagen results in different clinical and biochemical phenotypes. Human Genetics, 1996, 97, 324-329.	3.8	2
56	BglII RFLPs in the COL1A2 gene in the Finnish population. Human Genetics, 1988, 78, 109-109.	3.8	1
57	An anonymous genomic probe (pÎ»KP20.1) detects a multi-allelic locus on chromosome 19 (D19S25). Nucleic Acids Research, 1988, 16, 1643-1643.	14.5	1
58	Letter to the Editor Boning up on mutations: assessing the significance of candidate disease-causing DNA sequence variation. Genetics and Molecular Research, 2011, 10, 1518-1521.	0.2	1
59	Osteogenesis Imperfecta Genotypes and Genotypeâ€“Phenotype Relationships. , 2014, , 103-112.		1
60	Genetic counselling on brittle grounds: recurring osteogenesis imperfecta due to parental mosaicism for a dominant mutation. European Journal of Pediatrics, 1995, 154, 123-129.	2.7	1
61	An anonymous genomic probe (pÎ»KP20.1) detects a multi-allelic locus on chromosome 19 (DI9S25). Nucleic Acids Research, 1987, 15, 9105-9105.	14.5	0
62	Human type III collagen â€˜variantâ€™ is a cDNA cloning artefact. Nucleic Acids Research, 1988, 16, 11833-11833.	14.5	0
63	The Detection and Mapping of Point Mutations by RNase A Cleavage. , 1991, , 111-122.		0
64	Title is missing!. Trends in Biotechnology, 1997, 15, 378-379.	9.3	0
65	Southern Blotting. , 1998, , 2194-2198.		0
66	Hypophosphatasia: diagnostic application of linked DNA markers in the dominantly inherited adult form. Clinical Science, 1999, 97, 73.	4.3	0
67	Letters to the editors. Journal of Orthopaedic Research, 2006, 24, 1571-1574.	2.3	0
68	Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 790-792.	17.5	0
69	BRIF and Variant Databases. Nature Precedings, 2011, , .	0.1	0
70	A mini-library of sequenced human DNA fragments: linking bench experiments with informatics. Journal of Biological Education, 2012, 46, 193-198.	1.5	0
71	Mutations in COL1A1 Gene Change Dentin Nanostructure: A Response. Anatomical Record, 2018, 301, 1307-1308.	1.4	0
72	The comparison of the effectiveness of a modified conformation sensitive gel electrophoresis with denaturing high performance liquid chromatography. Iranian Biomedical Journal, 2008, 12, 109-14.	0.7	0

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73	Re: Long-term evaluation of anabolic and anti-resorptive agents in adults with familial osteoporosis due to pro205ala variant of the col1a1 gene. Osteoporosis International, 2022, , 1.	3.1	0