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	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
2	Dissecting the phenotypic variability of osteogenesis imperfecta. DMM Disease Models and Mechanisms, 2022, 15, .	2.4	16
3	Verifying nomenclature of DNA variants in submitted manuscripts: Guidance for journals. Human Mutation, 2021, 42, 3-7.	2.5	10
4	A common classification framework for histone sequence alterations in tumours: an expert consensus proposal. Journal of Pathology, 2021, 254, 109-120.	4.5	5
5	HUGO Gene Nomenclature Committee (HGNC) recommendations for the designation of gene fusions. Leukemia, 2021, 35, 3040-3043.	7.2	42
6	EMQN best practice guidelines for genetic testing in dystrophinopathies. European Journal of Human Genetics, 2020, 28, 1141-1159.	2.8	35
7	Mutations in COL1A1 Gene Change Dentin Nanostructure: A Response. Anatomical Record, 2018, 301, 1307-1308.	1.4	0
8	VariantValidator: Accurate validation, mapping, and formatting of sequence variation descriptions. Human Mutation, 2018, 39, 61-68.	2.5	105
9	hgvs: A Python package for manipulating sequence variants using HGVS nomenclature: 2018 Update. Human Mutation, 2018, 39, 1803-1813.	2.5	20
10	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
11	LSDBs and How They Have Evolved. Human Mutation, 2016, 37, 532-539.	2.5	6
12	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	2.5	1,194
13	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
14	Transcription factor SOHLH1 potentially associated with primary ovarian insufficiency. Fertility and Sterility, 2015, 103, 548-553.e5.	1.0	28
15	The Human Variome Project: ensuring the quality of DNA variant databases in inherited renal disease. Pediatric Nephrology, 2015, 30, 1893-1901.	1.7	2
16	Osteogenesis Imperfecta Genotypes and Genotypeâ€“Phenotype Relationships. , 2014, , 103-112.		1
17	Locus Reference Genomic: reference sequences for the reporting of clinically relevant sequence variants. Nucleic Acids Research, 2014, 42, D873-D878.	14.5	73
18	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

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19	Lack of association betweenESR1gene polymorphisms and premature ovarian failure in Serbian women. Climacteric, 2014, 17, 247-251.	2.4	9
20	Novel variants in the SOHLH2 gene are implicated in human premature ovarian failure. Fertility and Sterility, 2014, 101, 1104-1109.e6.	1.0	50
21	Variobox: Automatic Detection and Annotation of Human Genetic Variants. Human Mutation, 2014, 35, 202-207.	2.5	7
22	Quantifying the use of bioresources for promoting their sharing in scientific research. GigaScience, 2013, 2, 7.	6.4	38
23	Clinical utility gene card for: osteogenesis imperfecta. European Journal of Human Genetics, 2013, 21, 1-4.	2.8	68
24	A mini-library of sequenced human DNA fragments: linking bench experiments with informatics. Journal of Biological Education, 2012, 46, 193-198.	1.5	0
25	EMQN best practice guidelines for the laboratory diagnosis of osteogenesis imperfecta. European Journal of Human Genetics, 2012, 20, 11-19.	2.8	107
26	VarioML framework for comprehensive variation data representation and exchange. BMC Bioinformatics, 2012, 13, 254.	2.6	17
27	Curating gene variant databases (LSDBs): Toward a universal standard. Human Mutation, 2012, 33, 291-297.	2.5	41
28	Guidelines for establishing locus specific databases. Human Mutation, 2012, 33, 298-305.	2.5	48
29	Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 790-792.	17.5	0
30	BRIF and Variant Databases. Nature Precedings, 2011, , .	0.1	0
31	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
32	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
33	WAVe: web analysis of the variome. Human Mutation, 2011, 32, 729-734.	2.5	20
34	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
35	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	8.2	100
36	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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37	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
38	Letters to the editors. Journal of Orthopaedic Research, 2006, 24, 1571-1574.	2.3	0
39	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
40	Hypophosphatasia: diagnostic application of linked DNA markers in the dominantly inherited adult form. Clinical Science, 1999, 97, 73.	4.3	0
41	The Human Collagen Mutation Database 1998. Nucleic Acids Research, 1998, 26, 253-255.	14.5	198
42	Southern Blotting. , 1998, , 2194-2198.		0
43	The human type I collagen mutation database. Nucleic Acids Research, 1997, 25, 181-187.	14.5	282
44	Title is missing!. Trends in Biotechnology, 1997, 15, 378-379.	9.3	0
45	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
46	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
47	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
48	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
49	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
50	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
51	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
52	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
53	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
54	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

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55	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
56	The Detection and Mapping of Point Mutations by RNase A Cleavage. , 1991, , 111-122.		0
57	Length polymorphism in the threonine-glycine-encoding repeat region of theperiod gene inDrosophila. Journal of Molecular Evolution, 1991, 32, 238-246.	1.8	74
58	PCR detection of a COL1A1RsalRFLP. Nucleic Acids Research, 1991, 19, 3163-3163.	14.5	4
59	PCR detection of a 38 bp length variant in the COL1A2 gene. Nucleic Acids Research, 1990, 18, 5925-5925.	14.5	5
60	AHaeIII RFLP in COL1A1. Nucleic Acids Research, 1990, 18, 5926-5926.	14.5	3
61	BglII RFLPs in the COL1A2 gene in the Finnish population. Human Genetics, 1988, 78, 109-109.	3.8	1
62	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
63	Human type III collagen â€˜variantâ€™ is a cDNA cloning artefact. Nucleic Acids Research, 1988, 16, 11833-11833.	14.5	0
64	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
65	Human pro Î± 1(III) collagen: cDNA sequence for the 3â€² end. Nucleic Acids Research, 1988, 16, 2337-2337.	14.5	13
66	Collagen gene structure. Biochemical Society Transactions, 1988, 16, 661-663.	3.4	2
67	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
68	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
69	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
70	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
71	An RFLP associated with the human type III collagen gene (COL3A1). Nucleic Acids Research, 1985, 13, 4609-4609.	14.5	28
72	Isolation and partial sequence of recombinant plasmids containing human Î±-, Î²- and Î³-globin cDNA fragments. Nature, 1978, 273, 640-643.	27.8	78

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73	The divergence between human and baboon globin genes. Nucleic Acids and Protein Synthesis, 1976, 435, 76-81.	1.7	4