Peter Beighton

List of Publications by Year
in descending order

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64 papers	5,133 citations	236925 25 h-index	155660 55 g-index
65 all docs	65 docs citations	65 times ranked	4541 citing authors

#	Article	IF	Citations
1	Ehlers-Danlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	1,624
2	Bone Dysplasia Sclerosteosis Results from Loss of the SOST Gene Product, a Novel Cystine Knotâ€“Containing Protein. American Journal of Human Genetics, 2001, 68, 577-589.	6.2	912
3	Perlecan, the major proteoglycan of basement membranes, is altered in patients with Schwartz-Jampel syndrome (chondrodystrophic myotonia). Nature Genetics, 2000, 26, 480-483.	21.4	274
4	A 52-kb deletion in theSOST-MEOX1 intergenic region on 17q12-q21 is associated with van Buchem disease in the Dutch population. American Journal of Medical Genetics Part A, 2002, 110, 144-152.	2.4	270
5	Autosomal Dominant Craniometaphyseal Dysplasia Is Caused by Mutations in the Transmembrane Protein ANK. American Journal of Human Genetics, 2001, 68, 1321-1326.	6.2	177
6	Bone Mineral Density in Sclerosteosis; Affected Individuals and Gene Carriers. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 6392-6395.	3.6	162
7	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
8	A Mutation in the Variable Repeat Region of the Aggrecan Gene (AGC1) Causes a Form of Spondyloepiphyseal Dysplasia Associated with Severe, Premature Osteoarthritis. American Journal of Human Genetics, 2005, 77, 484-490.	6.2	137
9	The Clinical Features of Sclerosteosis. Annals of Internal Medicine, 1976, 84, 393.	3.9	115
10	A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. Human Molecular Genetics, 1994, 3, 915-918.	2.9	106
11	X-Linked Late-Onset Sensorineural Deafness Caused by a Deletion Involving OA1 and a Novel Gene Containing WD-40 Repeats. American Journal of Human Genetics, 1999, 64, 1604-1616.	6.2	97
12	Wormian bones in osteogenesis imperfecta and other disorders. Skeletal Radiology, 1982, 8, 35-38.	2.0	96
13	Localization of the Schwartzâ€“Jampel syndrome (SJS) locus to chromosome 1p34â€“p36.1 by homozygosity mapping. Human Molecular Genetics, 1995, 4, 1633-1636.	2.9	74
14	Sclerosteosis â€” An autosomal recessive disorder. Clinical Genetics, 1977, 11, 1-7.	2.0	68
15	Osteogenesis imperfecta with congenital joint contractures (Bruck Syndrome). Clinical Genetics, 1989, 36, 122-126.	2.0	60
16	The radiological spectrum of fibrodysplasia ossificans progressiva. Clinical Radiology, 1982, 33, 499-508.	1.1	52
17	Autosomal Dominant (Beukes) Premature Degenerative Osteoarthropathy of the Hip Joint Maps to an 11-cM Region on Chromosome 4q35. American Journal of Human Genetics, 1999, 64, 904-908.	6.2	49
18	Identification of a mutation in the ubiquitin-fold modifier 1-specific peptidase 2 gene, UFSP2, in an extended South African family with Beukes hip dysplasia. South African Medical Journal, 2015, 105, 558.	0.6	49

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19	Craniometaphyseal dysplasia â€variability of expression within a large family. Clinical Genetics, 1979, 15, 252-258.	2.0	48
20	An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. Human Molecular Genetics, 1995, 4, 1459-1462.	2.9	44
21	EhlersâEDanlos syndromes: Revised nosology, Villefranche, 1997. American Journal of Medical Genetics Part A, 1998, 77, 31-37.	2.4	40
22	Spondyloepiphyseal dysplasia, mild autosomal dominant type is not due to primary defects of type II collagen. American Journal of Medical Genetics Part A, 1990, 37, 272-276.	2.4	32
23	Childhood deafness in southern Africa. Journal of Laryngology and Otology, 1983, 97, 885-889.	0.8	31
24	Osteogenesis imperfects type III: An ancient mutation in Africa?. American Journal of Medical Genetics Part A, 1987, 27, 907-912.	2.4	29
25	Recessive Schwartz-Jampel syndrome (SJS): confirmation of linkage to chromosome 1p, evidence of genetic homogeneity and reduction of the SJS locus to a 3-cM interval. Human Genetics, 1996, 98, 380-385.	3.8	28
26	Pseudoxanthoma elasticum: Similar autosomal recessive subtype in Belgian and Afrikaner families. American Journal of Medical Genetics Part A, 1991, 38, 16-20.	2.4	24
27	The femoral hypoplasia â€unusual facies syndrome: A genetic entity?. Clinical Genetics, 1981, 20, 267-275.	2.0	23
28	Cataracts, alopecia, and sclerodactyly: A Previously apparently undescribed ectodermal dysplasia syndrome on the Island of Rodrigues. American Journal of Medical Genetics Part A, 1989, 32, 500-503.	2.4	22
29	Xâ€linked EhlersâEDanlos syndrome type V; the next generation. Clinical Genetics, 1985, 27, 472-478.	2.0	22
30	Namaqualand hip dysplasia: An autosomal dominant entity. American Journal of Medical Genetics Part A, 1984, 19, 161-169.	2.4	21
31	X-linked inheritance of ocular albinism with late-onset sensorineural deafness. American Journal of Medical Genetics Part A, 1984, 19, 797-803.	2.4	21
32	Piebaldism: an autonomous autosomal dominant entity. Clinical Genetics, 1991, 39, 330-337.	2.0	20
33	Familial rhizomelic dysplasia: Phenotypic variation or heterogeneity?. American Journal of Medical Genetics Part A, 1987, 26, 941-947.	2.4	19
34	Autosomal recessive inheritance in the mesomelic dwarfism of Campailla and Martinelli. Clinical Genetics, 1974, 5, 363-367.	2.0	18
35	Brachyolmia: An autosomal dominant form. American Journal of Medical Genetics Part A, 1994, 49, 308-312.	2.4	17
36	The Schwartz syndrome in Southern Africa. Clinical Genetics, 1973, 4, 548-555.	2.0	16

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37	Clinical and Genetic Aspects. , 1980, , 7-11.		16
38	Cutaneous manifestations of osteoectasia. Clinical and Experimental Dermatology, 1982, 7, 605-609.	1.3	15
39	Bruck syndrome: congenital joint contractures with bone fragility. Journal of Orthopaedic Science, 2005, 10, 641-646.	1.1	15
40	Osteoporosisâ€pseudoglioma syndrome. Clinical Genetics, 1986, 29, 263-263.	2.0	14
41	Fibrodysplasia ossificans progressiva (FOP) in South Africa: dental implications in 5 cases. Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics, 2011, 112, 11-18.	1.4	14
42	Brachydactylous dwarfs of Mseleni. American Journal of Medical Genetics Part A, 1993, 46, 636-640.	2.4	13
43	Autosomal recessive inheritance of osteogenesis imperfecta. Clinical Genetics, 1975, 8, 107-111.	2.0	13
44	Autosomal dominant (Beukes) premature degenerative osteoarthropathy of the hip joint unlinked to COL2A1. American Journal of Medical Genetics Part A, 1994, 53, 348-351.	2.4	12
45	Spondyloepiphyseal dysplasia in a cape town family: Linkage with the gene for type II collagen (COL2A1). American Journal of Medical Genetics Part A, 1992, 43, 833-838.	2.4	9
46	Marfan syndrome: a diagnostic dilemma. Clinical Genetics, 1990, 37, 417-422.	2.0	9
47	Osteochondrodystrophies with marked platyspondyly and distinctive peripheral anomalies. Pediatric Radiology, 1999, 29, 1-5.	2.0	8
48	Autosomal dominant inheritance of conductive deafness due to stapedial anomalies, external ear malformations and congenital facial palsy. Clinical Genetics, 1983, 23, 376-379.	2.0	8
49	Craniofacial manifestations in osteogenesis imperfecta type III in South Africa. BDJ Open, 2017, 3, 17021.	2.1	8
50	Rod-cone dystrophy, sensorineural deafness, and renal dysfunction: An autosomal recessive syndrome?. American Journal of Medical Genetics Part A, 1993, 47, 832-836.	2.4	7
51	Fibrodysplasia Ossificans Progressiva in South Africa. Journal of Clinical Rheumatology, 2011, 17, 37-41.	0.9	6
52	Heritable Hypermobility Syndromes. , 1999, , 147-177.		4
53	Osteochondrodysplasias in South Africa. , 1996, 63, 7-11.		3
54	Novel mutation in the BMPR1B gene (R486L) in a polish family and further delineation of the phenotypic features of BMPR1Bâ€Related brachydactyly. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 567-572.	1.6	3

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55	Renal dysfunction, rodâ€cone dystrophy, and sensorineural hearing loss caused by a mutation in RRM2B. Human Mutation, 2020, 41, 1871-1876.	2.5	3
56	Limb-girdle weakness in a marfanoid man: distinguishing calpainopathy from Becker's muscular dystrophy. Practical Neurology, 2015, 15, 152-154.	1.1	2
57	Skeletal Dysplasia Syndromes. , 2001, , 81-231.		1
58	SMD Kozlowski type caused by p.Arg594His substitution in TRPV4 reveals abnormal ossification and notochordal remnants in discs and vertebrae. European Journal of Medical Genetics, 2017, 60, 509-516.	1.3	1
59	Detecting genetic modifiers of spondyloepimetaphyseal dysplasia with joint laxity in the Caucasian Afrikaner community. Human Molecular Genetics, 2019, 28, 1053-1063.	2.9	1
60	Sclerosteosis. , 1980, , 109-122.		1
61	Skeletal Dysplasia Syndromes. , 1984, , 79-178.		1
62	Ultrasonic diagnosis of perinatal lethal hypophosphatasia. Ultrasound, 2013, 21, 132-136.	0.7	0
63	Heritable Hypermobility Syndromes. , 2012, , 151-189.		0
64	Craniometaphyseal Dysplasia. , 1980, , 55-65.		0