Richard M Pauli

List of Publications by Year
in descending order

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92 papers	4,459 citations	101496 36 h-index	114418 63 g-index
96 all docs	96 docs citations	96 times ranked	3904 citing authors

#	Article	IF	Citations
1	Mutations in the Transmembrane Natriuretic Peptide Receptor NPR-B Impair Skeletal Growth and Cause Acromesomelic Dysplasia, Type Maroteaux. American Journal of Human Genetics, 2004, 75, 27-34.	2.6	325
2	Achondroplasia: a comprehensive clinical review. Orphanet Journal of Rare Diseases, 2019, 14, 1.	1.2	292
3	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. Nature Genetics, 2011, 43, 303-305.	9.4	291
4	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	9.4	260
5	Apnea and sudden unexpected death in infants with achondroplasia. Journal of Pediatrics, 1984, 104, 342-348.	0.9	183
6	Stillbirth Classificationâ€"Developing an International Consensus for Research. Obstetrics and Gynecology, 2009, 114, 901-914.	1.2	168
7	Interstitial deletion of (17)(p11.2p11.2): Report of six additional patients with a new chromosome deletion syndrome. American Journal of Medical Genetics Part A, 1986, 24, 421-432.	2.4	127
8	Computerized tomography of the foramen magnum: Achondroplastic values compared to normal standards. American Journal of Medical Genetics Part A, 1985, 20, 355-360.	2.4	109
9	Natural history of rhizomelic chondrodysplasia punctata. American Journal of Medical Genetics Part A, 2003, 118A, 332-342.	2.4	104
10	Hajdu-Cheney syndrome: Evolution of phenotype and clinical problems. American Journal of Medical Genetics Part A, 2001, 100, 292-310.	2.4	102
11	Long-term survival in typical thanatophoric dysplasia type 1. American Journal of Medical Genetics Part A, 1997, 70, 427-436.	2.4	93
12	Ventricular noncompaction and distal chromosome 5q deletion. , 1999, 85, 419-423.		91
13	Familial agnathia-holoprosencephaly. American Journal of Medical Genetics Part A, 1983, 14, 677-698.	2.4	83
14	Wisconsin Stillbirth Service Program: II. Analysis of diagnoses and diagnostic categories in the first 1,000 referrals. American Journal of Medical Genetics Part A, 1994, 50, 135-153.	2.4	81
15	Autosomal Recessive Disorder Otospondylomegaepiphyseal Dysplasia Is Associated with Loss-of-Function Mutations in the COL11A2 Gene. American Journal of Human Genetics, 2000, 66, 368-377.	2.6	78
16	Homozygous achondroplasia with survival beyond infancy. American Journal of Medical Genetics Part A, 1983, 16, 459-473.	2.4	75
17	Obstructive Sleep Apnea in Children with Achondroplasia. Otolaryngology - Head and Neck Surgery, 1999, 120, 248-254.	1.1	75
18	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and WISP3 mutations in 63 affected individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 217-229.	0.7	74

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19	Mild hypophosphatasia mimicking severe osteogenesis imperfecta in utero: Bent but not broken. , 1999, 86, 434-438.		70
20	Prevention of Fixed, Angular Kyphosis in Achondroplasia. Journal of Pediatric Orthopaedics, 1997, 17, 726-733.	0.6	68
21	Temperament in Williams syndrome. American Journal of Medical Genetics Part A, 1990, 36, 345-352.	2.4	67
22	Wisconsin Stillbirth Service Program: I. Establishment and assessment of a community-based program for etiologic investigation of intrauterine deaths. American Journal of Medical Genetics Part A, 1994, 50, 116-134.	2.4	64
23	Major limb malformations following intrauterine exposure to ethanol: Two additional cases and literature review. Teratology, 1986, 33, 273-280.	1.7	57
24	Cranial nerve abnormalities in CHARGE association. American Journal of Medical Genetics Part A, 1993, 45, 751-757.	2.4	56
25	Imprinting Status of 11p15 Genes in Beckwithâ€“Wiedemann Syndrome Patients with CDKN1C Mutations. Genomics, 2001, 74, 370-376.	1.3	55
26	Spinocerebellar ataxia, hypogonadotropic hypogonadism, and choroidal dystrophy		

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37	Klippel-Feil anomaly with Sprengel anomaly, omovertebral bone, thumb abnormalities, and flexion-crease changes: Novel association or syndrome?. American Journal of Medical Genetics Part A, 2001, 101, 158-162.	2.4	37
38	Familial recurrence of terminal transverse defects of the arm. Clinical Genetics, 1985, 27, 555-563.	1.0	37
39	Title is missing!. Journal of Pediatric Orthopaedics, 1997, 17, 726-733.	0.6	36
40	Dominance and homozygosity in man. American Journal of Medical Genetics Part A, 1983, 16, 455-458.	2.4	34
41	Absence of predictable phenotypic expression in proximal 15q duplications. Clinical Genetics, 1991, 40, 194-201.	1.0	34
42	Limb deficiency and splenogonadal fusion. American Journal of Medical Genetics Part A, 1982, 13, 81-90.	2.4	33
43	Double heterozygosity in bone growth disorders: Four new observations and review. American Journal of Medical Genetics Part A, 2003, 121A, 193-208.	2.4	33
44	ERRORS IN THE PRENATAL DIAGNOSIS OF CHILDREN WITH ACHONDROPLASIA. , 1996, 16, 525-530.		31
45	Achondroplasia: Unexpected familial recurrence. American Journal of Medical Genetics Part A, 1984, 19, 245-250.	2.4	30
46	Autosomal dominant hypohidrotic ectodermal dysplasia in a large family. , 1997, 72, 462-467.		29
47	Arrayâ€based comparative genomic hybridization (aCGH) in the genetic evaluation of stillbirth. American Journal of Medical Genetics, Part A, 2009, 149A, 2437-2443.	0.7	29
48	Achondroplasia Natural History Study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. Genetics in Medicine, 2021, 23, 1498-1505.	1.1	29
49	Familial ovarian germ cell cancer: Report and review. , 1999, 84, 43-46.		27
50	Pelgerâ€“huet anomaly and a mild skeletal phenotype secondary to mutations in LBR. American Journal of Medical Genetics, Part A, 2013, 161, 2066-2073.	0.7	27
51	Atelosteogenesis type III: Long term survival, prenatal diagnosis, and evidence for dominant transmission. American Journal of Medical Genetics Part A, 1999, 83, 28-42.	2.4	26
52	Multicenter study of mortality in achondroplasia. American Journal of Medical Genetics, Part A, 2018, 176, 2359-2364.	0.7	25
53	Absence of correlation between infantile hypotonia and foramen magnum size in achondroplasia. American Journal of Medical Genetics Part A, 2001, 101, 40-45.	2.4	21
54	Spondylometaphyseal dysplasia with cone-rod dystrophy. American Journal of Medical Genetics Part A, 2004, 129A, 265-276.	2.4	19

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55	Beckwithâ€Wiedemann syndrome in adults: Observations from one family and recommendations for care. American Journal of Medical Genetics, Part A, 2008, 146A, 1707-1712.	0.7	19
56	Airway malacia in children with achondroplasia. American Journal of Medical Genetics, Part A, 2014, 164, 407-414.	0.7	19
57	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. American Journal of Medical Genetics Part A, 1984, 18, 311-320.	2.4	18
58	Standard curves of chest circumference in achondroplasia and the relationship of chest circumference to respiratory problems. , 1996, 62, 91-97.		18
59	Genotypeâ€“phenotype correlation in DTDST dysplasias: Atelosteogenesis type II and diastrophic dysplasia variant in one family. American Journal of Medical Genetics, Part A, 2010, 152A, 3043-3050.	0.7	18
60	Probable identity of Goltz syndrome and Van Allen-Myhre syndrome: Evidence from phenotypic evolution. American Journal of Medical Genetics Part A, 2002, 110, 370-379.	2.4	17
61	NovelCOL2A1Variant (c.619G>A, p.Gly207Arg) Manifesting as a Phenotype Similar to Progressive Pseudorheumatoid Dysplasia and Spondyloepiphyseal Dysplasia, Stanescu Type. Human Mutation, 2015, 36, 1004-1008.	1.1	17
62	Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2660-2669.	1.8	17
63	Optic atrophy, hearing loss, and peripheral neuropathy. American Journal of Medical Genetics Part A, 1989, 33, 61-65.	2.4	16
64	Smith-Lemli-Opitz syndrome: Thirty-year follow-up of â€œSâ€•of â€œRSHâ€•syndrome. , 1997, 68, 260-262.		16
65	An anadysplasiaâ€like, spontaneously remitting spondylometaphyseal dysplasia secondary to lamin B receptor (LBR) gene mutations: Further definition of the phenotypic heterogeneity of LBRâ€bone dysplasias. American Journal of Medical Genetics, Part A, 2015, 167, 159-163.	0.7	15
66	Jugular bulb dehiscence in achondroplasia. International Journal of Pediatric Otorhinolaryngology, 1999, 48, 169-174.	0.4	13
67	Karschâ€Neugebauer syndrome: split foot/split hand and congenital nystagmus. Clinical Genetics, 1985, 27, 97-101.	1.0	12
68	Growth in achondroplasia including stature, weight, weight-for-height and head circumference from CLARITY: achondroplasia natural history studyâ€”a multi-center retrospective cohort study of achondroplasia in the US. Orphanet Journal of Rare Diseases, 2021, 16, 522.	1.2	12
69	The natural histories of bone dysplasias in adultsâ€”Vignettes, fables and justâ€•so stories. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 309-321.	0.7	11
70	Neonatal death in cousins with trisomy 10q and monosomy 4p due to a familial translocation. Clinical Genetics, 1982, 22, 340-347.	1.0	11
71	Combined Phenotypes of Spondylometaphyseal Dysplasia-Kozlowski Type and Charcot-Marie-Tooth Disease Type 2C Secondary to a TRPV4 Pathogenic Variant. Molecular Syndromology, 2019, 10, 154-160.	0.3	11
72	Stillbirth: Fetal Disorders. Clinical Obstetrics and Gynecology, 2010, 53, 646-655.	0.6	10

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73	Does the clinical phenotype of mucolipidosis-IIIÎ³ differ from its Î±Î² counterpart?: supporting facts in a cohort of 18 patients. Clinical Dysmorphology, 2019, 28, 7-16.	0.1	10
74	Geleophysic dysplasia: 48 year clinical update with emphasis on cardiac care. American Journal of Medical Genetics, Part A, 2018, 176, 2237-2242.	0.7	8
75	Acanthosis nigricans in achondroplasia. American Journal of Medical Genetics, Part A, 2018, 176, 2630-2636.	0.7	7
76	Filippi syndrome: Report of three additional cases. , 1999, 87, 128-133.		6
77	<scp>CLARITY</scp>: Coâ€occurrences in achondroplasiaâ€"craniosynostosis, seizures, and decreased risk of diabetes mellitus. American Journal of Medical Genetics, Part A, 2021, 185, 1168-1174.	0.7	6
78	Frontonasal malformation and deletion of 22q11. , 1998, 75, 443-444.		5
79	Apparently benign craniocervical signs in achondroplasia: â€œneurologic leftoversâ€•identified through a retrospective dataset. Orphanet Journal of Rare Diseases, 2020, 15, 301.	1.2	5
80	Goldenhar association and cranial defects. American Journal of Medical Genetics Part A, 1983, 15, 177-179.	2.4	4
81	Unexpected familial recurrence in Angelman syndrome. , 1997, 70, 253-260.		4
82	Weight gain velocity in infants with achondroplasia. American Journal of Medical Genetics, Part A, 2020, 182, 146-149.	0.7	4
83	Craniocervical junction issues after infancy in achondroplasia. American Journal of Medical Genetics, Part A, 2021, 185, 182-189.	0.7	4
84	Achondroplasia Natural History Study (CLARITY): 60-year experience in cervicomedullary decompression in achondroplasia from four skeletal dysplasia centers. Journal of Neurosurgery: Pediatrics, 2021, , 1-7.	0.8	4
85	Typical achondroplasia secondary to a unique insertional variant of <scp>FGFR3</scp> with in vitro demonstration of its effect on <scp>FGFR3</scp> function. American Journal of Medical Genetics, Part A, 2021, 185, 798-805.	0.7	4
86	Validation of radiographic criteria for the diagnosis of Down syndrome in stillborn infants. American Journal of Medical Genetics Part A, 1997, 72, 347-350.	2.4	3
87	Letter to the editor: Response to two recent articles regarding achondroplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1099-1100.	0.7	2
88	Otolaryngology Utilization in Patients With Achondroplasia: Results From the CLARITY Study. Laryngoscope, 2021, , .	1.1	2
89	Reply to Dr. Lacombe. American Journal of Medical Genetics Part A, 1994, 49, 353-353.	2.4	1
90	Syringomyelia in hereditary multiple exostosis. American Journal of Medical Genetics, Part A, 2016, 170, 2956-2959.	0.7	1

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91	Achondroplasia is associated with increased occurrence of apparent lifeâ€threatening events. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 1842-1846.	0.7	1
92	Response to Drs. Hegde, Leung, and Robson. American Journal of Medical Genetics Part A, 1991, 39, 240-240.	2.4	0