

Richard M Pauli

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

93
papers

3,563
citations

34
h-index

58
g-index

96
ext. papers

4,060
ext. citations

4.7
avg, IF

5.31
L-index

#	Paper	IF	Citations
93	Mutations in the transmembrane natriuretic peptide receptor NPR-B impair skeletal growth and cause acromesomelic dysplasia, type Maroteaux. <i>American Journal of Human Genetics</i> , 2004 , 75, 27-34	11	279
92	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. <i>Nature Genetics</i> , 2011 , 43, 303-5	36.3	242
91	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. <i>Nature Genetics</i> , 1999 , 23, 94-8	36.3	227
90	Apnea and sudden unexpected death in infants with achondroplasia. <i>Journal of Pediatrics</i> , 1984 , 104, 342-8	3.6	159
89	Stillbirth classification--developing an international consensus for research: executive summary of a National Institute of Child Health and Human Development workshop. <i>Obstetrics and Gynecology</i> , 2009 , 114, 901-914	4.9	140
88	Achondroplasia: a comprehensive clinical review. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 1	4.2	131
87	Interstitial deletion of (17)(p11.2p11.2): report of six additional patients with a new chromosome deletion syndrome. <i>American Journal of Medical Genetics Part A</i> , 1986 , 24, 421-32		114
86	Computerized tomography of the foramen magnum: achondroplastic values compared to normal standards. <i>American Journal of Medical Genetics Part A</i> , 1985 , 20, 355-60		88
85	Hajdu-Cheney syndrome: evolution of phenotype and clinical problems. <i>American Journal of Medical Genetics Part A</i> , 2001 , 100, 292-310		84
84	Natural history of rhizomelic chondrodysplasia punctata. <i>American Journal of Medical Genetics Part A</i> , 2003 , 118A, 332-42		81
83	Familial agnathia-holoprosencephaly. <i>American Journal of Medical Genetics Part A</i> , 1983 , 14, 677-98		75
82	Ventricular noncompaction and distal chromosome 5q deletion 1999 , 85, 419-423		73
81	Wisconsin Stillbirth Service Program: II. Analysis of diagnoses and diagnostic categories in the first 1,000 referrals. <i>American Journal of Medical Genetics Part A</i> , 1994 , 50, 135-53		72
80	Long-term survival in typical thanatophoric dysplasia type 1 1997 , 70, 427-436		68
79	Homozygous achondroplasia with survival beyond infancy. <i>American Journal of Medical Genetics Part A</i> , 1983 , 16, 459-73		66
78	Autosomal recessive disorder otospondylomegaepiphyseal dysplasia is associated with loss-of-function mutations in the COL11A2 gene. <i>American Journal of Human Genetics</i> , 2000 , 66, 368-77	11	65
77	Mild hypophosphatasia mimicking severe osteogenesis imperfecta in utero: bent but not broken. <i>American Journal of Medical Genetics Part A</i> , 1999 , 86, 434-8		60

76	Obstructive sleep apnea in children with achondroplasia: surgical and anesthetic considerations. <i>Otolaryngology - Head and Neck Surgery</i> , 1999 , 120, 248-54	5.5	60
75	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): a review of clinical features, radiographic features, and WISP3 mutations in 63 affected individuals. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2012 , 160C, 217-29	3.1	59
74	Prevention of Fixed, Angular Kyphosis in Achondroplasia. <i>Journal of Pediatric Orthopaedics</i> , 1997 , 17, 726-733	2.4	58
73	Temperament in Williams syndrome. <i>American Journal of Medical Genetics Part A</i> , 1990 , 36, 345-52		57
72	Wisconsin Stillbirth Service Program: I. Establishment and assessment of a community-based program for etiologic investigation of intrauterine deaths. <i>American Journal of Medical Genetics Part A</i> , 1994 , 50, 116-34		56
71	Imprinting status of 11p15 genes in Beckwith-Wiedemann syndrome patients with CDKN1C mutations. <i>Genomics</i> , 2001 , 74, 370-6	4.3	53
70	Major limb malformations following intrauterine exposure to ethanol: two additional cases and literature review. <i>Teratology</i> , 1986 , 33, 273-80		51
69	Lower mesodermal defects: a common cause of fetal and early neonatal death. <i>American Journal of Medical Genetics Part A</i> , 1994 , 50, 154-72		49
68	Cranial nerve abnormalities in CHARGE association. <i>American Journal of Medical Genetics Part A</i> , 1993 , 45, 751-7		49
67	Spinocerebellar ataxia, hypogonadotropic hypogonadism, and choroidal dystrophy (Boucher-Neuhuser syndrome). <i>American Journal of Medical Genetics Part A</i> , 1989 , 33, 409-14		45
66	Tracheomalacia and bronchomalacia associated with Larsen syndrome. <i>Pediatric Pulmonology</i> , 1988 , 5, 55-9	3.5	44
65	Further evidence that arthrogryposis multiplex congenita in the human sometimes is caused by an intrauterine vascular accident. <i>Teratology</i> , 1992 , 45, 345-51		42
64	Mutations in PCYT1A, encoding a key regulator of phosphatidylcholine metabolism, cause spondylometaphyseal dysplasia with cone-rod dystrophy. <i>American Journal of Human Genetics</i> , 2014 , 94, 105-12	11	40
63	X-linked dominant chondrodysplasia punctata (CDPX2) caused by single gene mosaicism in a male. <i>American Journal of Medical Genetics Part A</i> , 2003 , 116A, 255-60		40
62	Head circumference of children with Down syndrome (0-36 months). <i>American Journal of Medical Genetics Part A</i> , 1992 , 42, 61-7		39
61	Mosaic isochromosome 12p. <i>American Journal of Medical Genetics Part A</i> , 1987 , 27, 291-4		35
60	Costs and consequences of comprehensive stillbirth assessment. <i>American Journal of Obstetrics and Gynecology</i> , 2002 , 186, 1027-34	6.4	34
59	Respiratory complications in children with spondyloepiphyseal dysplasia congenita. <i>Pediatric Pulmonology</i> , 1990 , 9, 49-54	3.5	34

58	Biophysical bases for delayed and aberrant motor development in young children with achondroplasia. <i>Journal of Developmental and Behavioral Pediatrics</i> , 1997 , 18, 143-50	2.4	32
57	Familial recurrence of terminal transverse defects of the arm. <i>Clinical Genetics</i> , 1985 , 27, 555-63	4	32
56	Klippel-Feil anomaly with Sprengel anomaly, omovertebral bone, thumb abnormalities, and flexion-crease changes: Novel association or syndrome?. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 158-162		32
55	Absence of predictable phenotypic expression in proximal 15q duplications. <i>Clinical Genetics</i> , 1991 , 40, 194-201	4	29
54	Double heterozygosity in bone growth disorders: four new observations and review. <i>American Journal of Medical Genetics Part A</i> , 2003 , 121A, 193-208		29
53	Achondroplasia: unexpected familial recurrence. <i>American Journal of Medical Genetics Part A</i> , 1984 , 19, 245-50		27
52	Familial ovarian germ cell cancer: report and review. <i>American Journal of Medical Genetics Part A</i> , 1999 , 84, 43-6		25
51	Errors in the prenatal diagnosis of children with achondroplasia. <i>Prenatal Diagnosis</i> , 1996 , 16, 525-30	3.2	25
50	Limb deficiency and splenogonadal fusion. <i>American Journal of Medical Genetics Part A</i> , 1982 , 13, 81-90		25
49	Pelger-huet anomaly and a mild skeletal phenotype secondary to mutations in LBR. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 2066-73	2.5	22
48	Array-based comparative genomic hybridization (aCGH) in the genetic evaluation of stillbirth. <i>American Journal of Medical Genetics, Part A</i> , 2009 , 149A, 2437-43	2.5	22
47	Atelosteogenesis type III: long term survival, prenatal diagnosis, and evidence for dominant transmission. <i>American Journal of Medical Genetics Part A</i> , 1999 , 83, 28-42		19
46	Spondylometaphyseal dysplasia with cone-rod dystrophy. <i>American Journal of Medical Genetics Part A</i> , 2004 , 129A, 265-76		18
45	. <i>Journal of Pediatric Orthopaedics</i> , 1997 , 17, 726-733	2.4	18
44	Absence of correlation between infantile hypotonia and foramen magnum size in achondroplasia. <i>American Journal of Medical Genetics Part A</i> , 2001 , 101, 40-5		17
43	Standard curves of chest circumference in achondroplasia and the relationship of chest circumference to respiratory problems. <i>American Journal of Medical Genetics Part A</i> , 1996 , 62, 91-7		17
42	Autosomal dominant hypohidrotic ectodermal dysplasia in a large family. <i>American Journal of Medical Genetics Part A</i> , 1997 , 72, 462-7		16
41	Optic atrophy, hearing loss, and peripheral neuropathy. <i>American Journal of Medical Genetics Part A</i> , 1989 , 33, 61-5		16

40	Novel COL2A1 variant (c.619G>A, p.Gly207Arg) manifesting as a phenotype similar to progressive pseudorheumatoid dysplasia and spondyloepiphyseal dysplasia, Stanescu type. <i>Human Mutation</i> , 2015 , 36, 1004-8	4.7	15
39	Probable identity of Goltz syndrome and Van Allen-Myhre syndrome: evidence from phenotypic evolution. <i>American Journal of Medical Genetics Part A</i> , 2002 , 110, 370-9		15
38	Airway malacia in children with achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 407-14	2.5	14
37	Genotype-phenotype correlation in DTDST dysplasias: Atelosteogenesis type II and diastrophic dysplasia variant in one family. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 3043-50	2.5	14
36	Beckwith-Wiedemann syndrome in adults: observations from one family and recommendations for care. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1707-12	2.5	14
35	Smith-Lemli-Opitz syndrome: thirty-year follow-up of "S" of "RSH" syndrome. <i>American Journal of Medical Genetics Part A</i> , 1997 , 68, 260-2		13
34	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. <i>American Journal of Medical Genetics Part A</i> , 1984 , 18, 311-20		13
33	Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018 , 103, 2660-2669	5.6	12
32	An anadysplasia-like, spontaneously remitting spondylometaphyseal dysplasia secondary to lamin B receptor (LBR) gene mutations: further definition of the phenotypic heterogeneity of LBR-bone dysplasias. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 159-63	2.5	11
31	Neonatal death in cousins with trisomy 10q and monosomy 4p due to a familial translocation. <i>Clinical Genetics</i> , 1982 , 22, 340-7	4	11
30	Stillbirth: fetal disorders. <i>Clinical Obstetrics and Gynecology</i> , 2010 , 53, 646-55	1.7	10
29	The natural histories of bone dysplasias in adults--vignettes, fables and just-so stories. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2007 , 145C, 309-21	3.1	10
28	Multicenter study of mortality in achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2359-2364	2.5	10
27	Karsch-Neugebauer syndrome: split foot/split hand and congenital nystagmus. <i>Clinical Genetics</i> , 1985 , 27, 97-101	4	9
26	Jugular bulb dehiscence in achondroplasia. <i>International Journal of Pediatric Otorhinolaryngology</i> , 1999 , 48, 169-74	1.7	9
25	Combined Phenotypes of Spondylometaphyseal Dysplasia-Kozlowski Type and Charcot-Marie-Tooth Disease Type 2C Secondary to a TRPV4 Pathogenic Variant. <i>Molecular Syndromology</i> , 2019 , 10, 154-160	1.5	7
24	Does the clinical phenotype of mucopolisidosis-III differ from its counterpart?: supporting facts in a cohort of 18 patients. <i>Clinical Dysmorphology</i> , 2019 , 28, 7-16	0.9	6
23	Achondroplasia Natural History Study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. <i>Genetics in Medicine</i> , 2021 , 23, 1498-1505	8.1	5

22	Acanthosis nigricans in achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2630-2636	5
21	Geleophysic dysplasia: 48 year clinical update with emphasis on cardiac care. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2237-2242	2.5 5
20	Frontonasal malformation and deletion of 22q11 1998 , 75, 443-444	4
19	Filippi syndrome: Report of three additional cases 1999 , 87, 128-133	4
18	Goldenhar association and cranial defects. <i>American Journal of Medical Genetics Part A</i> , 1983 , 15, 177-9	4
17	Achondroplasia 2010 , 17-37	3
16	Unexpected familial recurrence in Angelman syndrome. <i>American Journal of Medical Genetics Part A</i> , 1997 , 70, 253-60	3
15	Validation of radiographic criteria for the diagnosis of Down syndrome in stillborn infants. <i>American Journal of Medical Genetics Part A</i> , 1997 , 72, 347-50	3
14	Letter to the editor: Response to two recent articles regarding achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170A, 1099-100	2.5 2
13	Apparently benign craniocervical signs in achondroplasia: "neurologic leftovers" identified through a retrospective dataset. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 301	4.2 1
12	Reply to Dr. Lacombe. <i>American Journal of Medical Genetics Part A</i> , 1994 , 49, 353-353	1
11	ACHONDROPLASIA 2021 , 9-30	1
10	Weight gain velocity in infants with achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 146-149	2.5 1
9	Syringomyelia in hereditary multiple exostosis. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2956-2959	2.5 1
8	Craniocervical junction issues after infancy in achondroplasia. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 182-189	2.5 1
7	Achondroplasia is associated with increased occurrence of apparent life-threatening events. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2021 , 110, 1842-1846	3.1 1
6	Achondroplasia Natural History Study (CLARITY): 60-year experience in cervicomedullary decompression in achondroplasia from four skeletal dysplasia centers. <i>Journal of Neurosurgery: Pediatrics</i> , 2021 , 1-7	2.1 1
5	Achondroplasia	1

4	Typical achondroplasia secondary to a unique insertional variant of FGFR3 with in vitro demonstration of its effect on FGFR3 function. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 798-805	2.5	1
3	CLARITY: Co-occurrences in achondroplasia-craniosynostosis, seizures, and decreased risk of diabetes mellitus. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1168-1174	2.5	0
2	Growth in achondroplasia including stature, weight, weight-for-height and head circumference from CLARITY: achondroplasia natural history study-a multi-center retrospective cohort study of achondroplasia in the US.. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 522	4.2	0
1	Response to Drs. Hegde, Leung, and Robson. <i>American Journal of Medical Genetics Part A</i> , 1991 , 39, 240-240		