Richard M Pauli

List of Publications by Year in Descending Order

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	ACHONDROPLASIA **2021**, 9-30		1
92	Achondroplasia Natural History Study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. *Genetics in Medicine*, **2021**, 23, 1498-1505	8.1	5
91	Craniocervical junction issues after infancy in achondroplasia. *American Journal of Medical Genetics, Part A*, **2021**, 185, 182-189	2.5	1
90	CLARITY: Co-occurrences in achondroplasia-craniosynostosis, seizures, and decreased risk of diabetes mellitus. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1168-1174	2.5	0
89	Achondroplasia is associated with increased occurrence of apparent life-threatening events. *Acta Paediatrica, International Journal of Paediatrics*, **2021**, 110, 1842-1846	3.1	1
88	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	2.1	1
87	Typical achondroplasia secondary to a unique insertional variant of FGFR3 with in vitro demonstration of its effect on FGFR3 function. *American Journal of Medical Genetics, Part A*, **2021**, 185, 798-805	2.5	1
86	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.. *Orphanet Journal of Rare Diseases*, **2021**, 16, 522	4.2	0
85	Apparently benign craniocervical signs in achondroplasia: "neurologic leftovers" identified through a retrospective dataset. *Orphanet Journal of Rare Diseases*, **2020**, 15, 301	4.2	1
84	Weight gain velocity in infants with achondroplasia. *American Journal of Medical Genetics, Part A*, **2020**, 182, 146-149	2.5	1
83	Does the clinical phenotype of mucolipidosis-III differ from its counterpart?: supporting facts in a cohort of 18 patients. *Clinical Dysmorphology*, **2019**, 28, 7-16	0.9	6
82	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	1.5	7
81	Achondroplasia: a comprehensive clinical review. *Orphanet Journal of Rare Diseases*, **2019**, 14, 1	4.2	131
80	Progression of Mineral Ion Abnormalities in Patients With Jansen Metaphyseal Chondrodysplasia. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 2660-2669	5.6	12
79	Multicenter study of mortality in achondroplasia. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2359-2364	2.5	10
78	Acanthosis nigricans in achondroplasia. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2630-2636	2.5	5
77	Geleophysic dysplasia: 48 year clinical update with emphasis on cardiac care. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2237-2242	2.5	5

76	Letter to the editor: Response to two recent articles regarding achondroplasia. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 1099-100	2.5	2
75	Syringomyelia in hereditary multiple exostosis. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2956-2959	2.5	1
74	Novel COL2A1 variant (c.619G>A, p.Gly207Arg) manifesting as a phenotype similar to progressive pseudorheumatoid dysplasia and spondyloepiphyseal dysplasia, Stanescu type. *Human Mutation*, **2015**, 36, 1004-8	4.7	15
73	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. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 159-63	2.5	11
72	Airway malacia in children with achondroplasia. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 407-14	2.5	14
71	Mutations in PCYT1A, encoding a key regulator of phosphatidylcholine metabolism, cause spondylometaphyseal dysplasia with cone-rod dystrophy. *American Journal of Human Genetics*, **2014**, 94, 105-12	11	40
70	Pelger-huet anomaly and a mild skeletal phenotype secondary to mutations in LBR. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2066-73	2.5	22
69	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, 217-29	3.1	59
68	Mutations in NOTCH2 cause Hajdu-Cheney syndrome, a disorder of severe and progressive bone loss. *Nature Genetics*, **2011**, 43, 303-5	36.3	242
67	Stillbirth: fetal disorders. *Clinical Obstetrics and Gynecology*, **2010**, 53, 646-55	1.7	10
66	Achondroplasia **2010**, 17-37		3
65	Genotype-phenotype correlation in DTDST dysplasias: Atelosteogenesis type II and diastrophic dysplasia variant in one family. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 3043-50	2.5	14
64	Array-based comparative genomic hybridization (aCGH) in the genetic evaluation of stillbirth. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 2437-43	2.5	22
63	Stillbirth classification--developing an international consensus for research: executive summary of a National Institute of Child Health and Human Development workshop. *Obstetrics and Gynecology*, **2009**, 114, 901-914	4.9	140
62	Beckwith-Wiedemann syndrome in adults: observations from one family and recommendations for care. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1707-12	2.5	14
61	The natural histories of bone dysplasias in adults--vignettes, fables and just-so stories. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2007**, 145C, 309-21	3.1	10
60	Spondylometaphyseal dysplasia with cone-rod dystrophy. *American Journal of Medical Genetics Part A*, **2004**, 129A, 265-76		18
59	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	11	279

58	X-linked dominant chondrodysplasia punctata (CDPX2) caused by single gene mosaicism in a male. *American Journal of Medical Genetics Part A*, **2003**, 116A, 255-60		40
57	Natural history of rhizomelic chondrodysplasia punctata. *American Journal of Medical Genetics Part A*, **2003**, 118A, 332-42		81
56	Double heterozygosity in bone growth disorders: four new observations and review. *American Journal of Medical Genetics Part A*, **2003**, 121A, 193-208		29
55	Costs and consequences of comprehensive stillbirth assessment. *American Journal of Obstetrics and Gynecology*, **2002**, 186, 1027-34	6.4	34
54	Probable identity of Goltz syndrome and Van Allen-Myhre syndrome: evidence from phenotypic evolution. *American Journal of Medical Genetics Part A*, **2002**, 110, 370-9		15
53	Hajdu--Cheney syndrome: evolution of phenotype and clinical problems. *American Journal of Medical Genetics Part A*, **2001**, 100, 292-310		84
52	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		32
51	Absence of correlation between infantile hypotonia and foramen magnum size in achondroplasia. *American Journal of Medical Genetics Part A*, **2001**, 101, 40-5		17
50	Imprinting status of 11p15 genes in Beckwith-Wiedemann syndrome patients with CDKN1C mutations. *Genomics*, **2001**, 74, 370-6	4.3	53
49	Autosomal recessive disorder otospondylomegaepiphyseal dysplasia is associated with loss-of-function mutations in the COL11A2 gene. *American Journal of Human Genetics*, **2000**, 66, 368-77	11	65
48	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. *Nature Genetics*, **1999**, 23, 94-8	36.3	227
47	Atelosteogenesis type III: long term survival, prenatal diagnosis, and evidence for dominant transmission. *American Journal of Medical Genetics Part A*, **1999**, 83, 28-42		19
46	Familial ovarian germ cell cancer: report and review. *American Journal of Medical Genetics Part A*, **1999**, 84, 43-6		25
45	Ventricular noncompaction and distal chromosome 5q deletion **1999**, 85, 419-423		73
44	Mild hypophosphatasia mimicking severe osteogenesis imperfecta in utero: bent but not broken. *American Journal of Medical Genetics Part A*, **1999**, 86, 434-8		60
43	Filippi syndrome: Report of three additional cases **1999**, 87, 128-133		4
42	Jugular bulb dehiscence in achondroplasia. *International Journal of Pediatric Otorhinolaryngology*, **1999**, 48, 169-74	1.7	9
41	Obstructive sleep apnea in children with achondroplasia: surgical and anesthetic considerations. *Otolaryngology - Head and Neck Surgery*, **1999**, 120, 248-54	5.5	60

40	Frontonasal malformation and deletion of 22q11 **1998**, 75, 443-444		4
39	Biophysical bases for delayed and aberrant motor development in young children with achondroplasia. *Journal of Developmental and Behavioral Pediatrics*, **1997**, 18, 143-50	2.4	32
38	Smith-Lemli-Opitz syndrome: thirty-year follow-up of "S" of "RSH" syndrome. *American Journal of Medical Genetics Part A*, **1997**, 68, 260-2		13
37	Unexpected familial recurrence in Angelman syndrome. *American Journal of Medical Genetics Part A* , **1997**, 70, 253-60		3
36	Long-term survival in typical thanatophoric dysplasia type 1 **1997**, 70, 427-436		68
35	Validation of radiographic criteria for the diagnosis of Down syndrome in stillborn infants. *American Journal of Medical Genetics Part A*, **1997**, 72, 347-50		3
34	Autosomal dominant hypohidrotic ectodermal dysplasia in a large family. *American Journal of Medical Genetics Part A*, **1997**, 72, 462-7		16
33	. *Journal of Pediatric Orthopaedics*, **1997**, 17, 726-733	2.4	18
32	Prevention of Fixed, Angular Kyphosis in Achondroplasia. *Journal of Pediatric Orthopaedics*, **1997**, 17, 726-733	2.4	58
31	Standard curves of chest circumference in achondroplasia and the relationship of chest circumference to respiratory problems. *American Journal of Medical Genetics Part A*, **1996**, 62, 91-7		17
30	Errors in the prenatal diagnosis of children with achondroplasia. *Prenatal Diagnosis*, **1996**, 16, 525-30	3.2	25
29	Reply to Dr. Lacombe. *American Journal of Medical Genetics Part A*, **1994**, 49, 353-353		1
28	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, 116-34		56
27	Lower mesodermal defects: a common cause of fetal and early neonatal death. *American Journal of Medical Genetics Part A*, **1994**, 50, 154-72		49
26	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, 135-53		72
25	Cranial nerve abnormalities in CHARGE association. *American Journal of Medical Genetics Part A*, **1993**, 45, 751-7		49
24	Further evidence that arthrogryposis multiplex congenita in the human sometimes is caused by an intrauterine vascular accident. *Teratology*, **1992**, 45, 345-51		42
23	Head circumference of children with Down syndrome (0-36 months). *American Journal of Medical Genetics Part A*, **1992**, 42, 61-7		39

22	Absence of predictable phenotypic expression in proximal 15q duplications. *Clinical Genetics*, **1991**, 40, 194-201	4	29
21	Response to Drs. Hegde, Leung, and Robson. *American Journal of Medical Genetics Part A*, **1991**, 39, 240-240		
20	Respiratory complications in children with spondyloepiphyseal dysplasia congenita. *Pediatric Pulmonology*, **1990**, 9, 49-54	3.5	34
19	Temperament in Williams syndrome. *American Journal of Medical Genetics Part A*, **1990**, 36, 345-52		57
18	Optic atrophy, hearing loss, and peripheral neuropathy. *American Journal of Medical Genetics Part A*, **1989**, 33, 61-5		16
17	Spinocerebellar ataxia, hypogonadotropic hypogonadism, and choroidal dystrophy (Boucher-Neuhäuser syndrome). *American Journal of Medical Genetics Part A*, **1989**, 33, 409-14		45
16	Tracheomalacia and bronchomalacia associated with Larsen syndrome. *Pediatric Pulmonology*, **1988**, 5, 55-9	3.5	44
15	Mosaic isochromosome 12p. *American Journal of Medical Genetics Part A*, **1987**, 27, 291-4		35
14	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, 421-32		114
13	Major limb malformations following intrauterine exposure to ethanol: two additional cases and literature review. *Teratology*, **1986**, 33, 273-80		51
12	Karsch-Neugebauer syndrome: split foot/split hand and congenital nystagmus. *Clinical Genetics*, **1985**, 27, 97-101	4	9
11	Familial recurrence of terminal transverse defects of the arm. *Clinical Genetics*, **1985**, 27, 555-63	4	32
10	Computerized tomography of the foramen magnum: achondroplastic values compared to normal standards. *American Journal of Medical Genetics Part A*, **1985**, 20, 355-60		88
9	Genetic heterogeneity in spondyloepiphyseal dysplasia congenita. *American Journal of Medical Genetics Part A*, **1984**, 18, 311-20		13
8	Achondroplasia: unexpected familial recurrence. *American Journal of Medical Genetics Part A*, **1984**, 19, 245-50		27
7	Apnea and sudden unexpected death in infants with achondroplasia. *Journal of Pediatrics*, **1984**, 104, 342-8	3.6	159
6	Familial agnathia-holoprosencephaly. *American Journal of Medical Genetics Part A*, **1983**, 14, 677-98		75
5	Goldenhar association and cranial defects. *American Journal of Medical Genetics Part A*, **1983**, 15, 177-9		4

4	Homozygous achondroplasia with survival beyond infancy. *American Journal of Medical Genetics Part A*, **1983**, 16, 459-73		66
3	Neonatal death in cousins with trisomy 10q and monosomy 4p due to a familial translocation. *Clinical Genetics*, **1982**, 22, 340-7	4	11
2	Limb deficiency and splenogonadal fusion. *American Journal of Medical Genetics Part A*, **1982**, 13, 81-90		25
1	Achondroplasia		1