Daniel H Cohn

List of Publications by Year in Descending Order

67 papers	4,207 citations	31 h-index	64 g-index
67 ext. papers	4,870 ext. citations	11.4 avg, IF	4.44 L-index

#	Paper	IF	Citations
67	Mutations in GRK2 cause Jeune syndrome by impairing Hedgehog and canonical Wnt signaling. *EMBO Molecular Medicine*, **2020**, 12, e11739	12	7
66	Biallelic mutations in LAMA5 disrupts a skeletal noncanonical focal adhesion pathway and produces a distinct bent bone dysplasia. *EBioMedicine*, **2020**, 62, 103075	8.8	3
65	Nosology and classification of genetic skeletal disorders: 2019 revision. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2393-2419	2.5	216
64	The 2 chain of type IX collagen is essential for type IX collagen biosynthesis. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1672-1677	2.5	1
63	Dominant-negative SOX9 mutations in campomelic dysplasia. *Human Mutation*, **2019**, 40, 2344-2352	4.7	8
62	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
61	Expanding the genetic architecture and phenotypic spectrum in the skeletal ciliopathies. *Human Mutation*, **2018**, 39, 152-166	4.7	51
60	Autosomal recessive Stickler syndrome resulting from a COL9A3 mutation. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2887-2891	2.5	14
59	The PTH/PTHrP-SIK3 pathway affects skeletogenesis through altered mTOR signaling. *Science Translational Medicine*, **2018**, 10,	17.5	21
58	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGF signaling and cause autosomal dominant spondylocarpotarsal synostosis. *Scientific Reports*, **2017**, 7, 41803	4.9	17
57	A Chaperone Complex Formed by HSP47, FKBP65, and BiP Modulates Telopeptide Lysyl Hydroxylation of Type I Procollagen. *Journal of Bone and Mineral Research*, **2017**, 32, 1309-1319	6.3	41
56	Mutations in IFT-A satellite core component genes and produce short rib polydactyly syndrome with distinctive campomelia. *Cilia*, **2017**, 6, 7	5.5	16
55	Genes uniquely expressed in human growth plate chondrocytes uncover a distinct regulatory network. *BMC Genomics*, **2017**, 18, 983	4.5	7
54	MED resulting from recessively inherited mutations in the gene encoding calcium-activated nucleotidase CANT1. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2415-2421	2.5	14
53	Somatic mosaicism for a lethal TRPV4 mutation results in non-lethal metatropic dysplasia. *American Journal of Medical Genetics, Part A*, **2016**, 170, 3298-3302	2.5	6
52	Destabilization of the IFT-B cilia core complex due to mutations in IFT81 causes a Spectrum of Short-Rib Polydactyly Syndrome. *Scientific Reports*, **2016**, 6, 34232	4.9	30
51	Altered mRNA Splicing, Chondrocyte Gene Expression and Abnormal Skeletal Development due to SF3B4 Mutations in Rodriguez Acrofacial Dysostosis. *PLoS Genetics*, **2016**, 12, e1006307	6	22

50	Clinical and radiographic delineation of Bent Bone Dysplasia-FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angel-shaped Phalanges. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2652-61	2.5	6
49	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive WNT1 mutations. *Journal of Medical Genetics*, **2016**, 53, 427-30	5.8	30
48	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. *Nature Genetics*, **2016**, 48, 648-56	36.3	78
47	An inactivating mutation in intestinal cell kinase, ICK, impairs hedgehog signalling and causes short rib-polydactyly syndrome. *Human Molecular Genetics*, **2016**, 25, 3998-4011	5.6	31
46	IFT52 mutations destabilize anterograde complex assembly, disrupt ciliogenesis and result in short rib polydactyly syndrome. *Human Molecular Genetics*, **2016**, 25, 4012-4020	5.6	25
45	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. *Nature Communications*, **2015**, 6, 7092	17.4	55
44	HSP47 and FKBP65 cooperate in the synthesis of type I procollagen. *Human Molecular Genetics*, **2015**, 24, 1918-28	5.6	39
43	A second locus for Schneckenbecken dysplasia identified by a mutation in the gene encoding inositol polyphosphate phosphatase-like 1 (INPPL1). *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2470-3	2.5	8
42	Mice expressing mutant Trpv4 recapitulate the human TRPV4 disorders. *Journal of Bone and Mineral Research*, **2014**, 29, 1815-1822	6.3	15
41	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 213-219	1.8	14
40	Follistatin in chondrocytes: the link between TRPV4 channelopathies and skeletal malformations. *FASEB Journal*, **2014**, 28, 2525-37	0.9	24
39	Opsismodysplasia resulting from an insertion mutation in the SH2 domain, which destabilizes INPPL1. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2407-11	2.5	10
38	WDR34 mutations that cause short-rib polydactyly syndrome type III/severe asphyxiating thoracic dysplasia reveal a role for the NF-B pathway in cilia. *American Journal of Human Genetics*, **2013**, 93, 926-31	11	66
37	Whole-genome analysis reveals that mutations in inositol polyphosphate phosphatase-like 1 cause opsismodysplasia. *American Journal of Human Genetics*, **2013**, 92, 137-43	11	46
36	WNT1 mutations in early-onset osteoporosis and osteogenesis imperfecta. *New England Journal of Medicine*, **2013**, 368, 1809-16	59.2	253
35	Dominant and recessive forms of fibrochondrogenesis resulting from mutations at a second locus, COL11A2. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 309-14	2.5	14
34	Exome sequencing identifies PDE4D mutations in acrodysostosis. *American Journal of Human Genetics*, **2012**, 90, 746-51	11	114
33	Fibrochondrogenesis results from mutations in the COL11A1 type XI collagen gene. *American Journal of Human Genetics*, **2010**, 87, 708-12	11	56

32	Dominant TRPV4 mutations in nonlethal and lethal metatropic dysplasia. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1169-77	2.5	71
31	A recessive skeletal dysplasia, SEMD aggrecan type, results from a missense mutation affecting the C-type lectin domain of aggrecan. *American Journal of Human Genetics*, **2009**, 84, 72-9	11	94
30	Mutations in the gene encoding the calcium-permeable ion channel TRPV4 produce spondylometaphyseal dysplasia, Kozlowski type and metatropic dysplasia. *American Journal of Human Genetics*, **2009**, 84, 307-15	11	148
29	Ciliary abnormalities due to defects in the retrograde transport protein DYNC2H1 in short-rib polydactyly syndrome. *American Journal of Human Genetics*, **2009**, 84, 542-9	11	131
28	Gain-of-function mutations in TRPV4 cause autosomal dominant brachyolmia. *Nature Genetics*, **2008**, 40, 999-1003	36.3	295
27	Nucleotide-sugar transporter SLC35D1 is critical to chondroitin sulfate synthesis in cartilage and skeletal development in mouse and human. *Nature Medicine*, **2007**, 13, 1363-7	50.5	91
26	Cartilage-selective genes identified in genome-scale analysis of non-cartilage and cartilage gene expression. *BMC Genomics*, **2007**, 8, 165	4.5	17
25	MED, COMP, multilayered and NEIN: an overview of multiple epiphyseal dysplasia. *Pediatric Radiology*, **2005**, 35, 116-23	2.8	43
24	A transcriptional profile of human fetal cartilage. *Matrix Biology*, **2004**, 23, 299-307	11.4	17
23	Mental retardation and abnormal skeletal development (Dyggve-Melchior-Clausen dysplasia) due to mutations in a novel, evolutionarily conserved gene. *American Journal of Human Genetics*, **2003**, 72, 419-28	11	43
22	Analysis of clones from a human cartilage cDNA library provides insight into chondrocyte gene expression and identifies novel candidate genes for the osteochondrodysplasias. *Molecular Genetics and Metabolism*, **2003**, 79, 34-42	3.7	10
21	Defects in extracellular matrix structural proteins in the osteochondrodysplasias. *Novartis Foundation Symposium*, **2001**, 232, 195-210; discussion 210-2		7
20	Double heterozygosity for pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. *American Journal of Medical Genetics Part A*, **2001**, 104, 140-6		37
19	Widely distributed mutations in the COL2A1 gene produce achondrogenesis type II/hypochondrogenesis. *American Journal of Medical Genetics Part A*, **2000**, 92, 95-100		52
18	Rapid determination of COL2A1 mutations in individuals with Stickler syndrome: analysis of potential premature termination codons. *American Journal of Medical Genetics Part A*, **2000**, 94, 141-8		24
17	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. *Nature Genetics*, **1999**, 21, 302-4	36.3	285
16	Small deletions in the type II collagen triple helix produce Kniest dysplasia **1999**, 85, 105-112		47
15	Small deletions in the type II collagen triple helix produce Kniest dysplasia **1999**, 85, 105		1

14	Identification of five novel mutations in cartilage oligomeric matrix protein gene in pseudoachondroplasia and multiple epiphyseal dysplasia. *Human Mutation*, **1998**, Suppl 1, S10-7	4.7	29
13	Distinct, autosomal recessive form of spondyloepimetaphyseal dysplasia segregating in an inbred Pakistani kindred. *American Journal of Medical Genetics Part A*, **1998**, 78, 468-473		44
12	Correlation of linkage data with phenotype in eight families with Stickler syndrome. *American Journal of Medical Genetics Part A*, **1998**, 80, 121-127		40
11	Radiographic and morphologic findings in a previously undescribed type of mesomelic dysplasia resembling atelosteogenesis type II. *American Journal of Medical Genetics Part A*, **1998**, 80, 247-251		11
10	Diverse mutations in the gene for cartilage oligomeric matrix protein in the pseudoachondroplasia-multiple epiphyseal dysplasia disease spectrum. *American Journal of Human Genetics*, **1998**, 62, 311-9	11	148
9	Clinical, morphological, and biochemical phenotype of a new case of Ehlers-Danlos syndrome type VIIC. *American Journal of Medical Genetics Part A*, **1997**, 68, 25-8		24
8	Sulfate transport in chondrodysplasia. *Annals of the New York Academy of Sciences*, **1996**, 785, 131-6	6.5	14
7	Achondrogenesis type IB is caused by mutations in the diastrophic dysplasia sulphate transporter gene. *Nature Genetics*, **1996**, 12, 100-2	36.3	189
6	Thanatophoric dysplasia (types I and II) caused by distinct mutations in fibroblast growth factor receptor 3. *Nature Genetics*, **1995**, 9, 321-8	36.3	524
5	Dominant mutations in the type II collagen gene, COL2A1, produce spondyloepimetaphyseal dysplasia, Strudwick type. *Nature Genetics*, **1995**, 11, 87-9	36.3	76
4	Homology-mediated recombination between type I collagen gene exons results in an internal tandem duplication and lethal osteogenesis imperfecta. *Human Mutation*, **1993**, 2, 21-7	4.7	10
3	First-trimester prenatal diagnosis of osteogenesis imperfecta type II by DNA analysis and sonography. *Prenatal Diagnosis*, **1993**, 13, 589-96	3.2	30
2	A type X collagen mutation causes Schmid metaphyseal chondrodysplasia. *Nature Genetics*, **1993**, 5, 79-82	36.3	219
1	Recurrence of lethal osteogenesis imperfecta due to parental mosaicism for a mutation in the COL1A2 gene of type I collagen. The mosaic parent exhibits phenotypic features of a mild form of the disease. *Human Mutation*, **1992**, 1, 47-54	4.7	68