Deborah Krakow

List of Publications by Year
in descending order

159 papers	9,720 citations	46984 47 h-index	40954 93 g-index
166 all docs	166 docs citations	166 times ranked	10711 citing authors

#	Article	IF	Citations
1	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
2	Intervertebral disc degeneration is rescued by TGFÎ²/BMP signaling modulation in an ex vivo filamin B mouse model. Bone Research, 2022, 10, 37.	5.4	4
3	Emergency department use among postpartum women with mental health disorders. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100269.	1.3	12
4	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	1.1	16
5	Revisiting Skeletal Dysplasias in the Newborn. NeoReviews, 2021, 22, e216-e229.	0.4	0
6	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	1.1	10
7	Innovations in MD-only physician-scientist training: experiences from the Burroughs Wellcome Fund physician-scientist institutional award initiative. Journal of Clinical Investigation, 2021, 131, .	3.9	4
8	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
9	An RNA aptamer restores defective bone growth in FGFR3-related skeletal dysplasia in mice. Science Translational Medicine, 2021, 13, .	5.8	20
10	Osteogenesis imperfecta tooth level phenotype analysis: Cross-sectional study. Bone, 2021, 147, 115917.	1.4	7
11	Localized chondro-ossification underlies joint dysfunction and motor deficits in the Fkbp10 mouse model of osteogenesis imperfecta. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	3
12	Pregnancy in women with osteogenesis imperfecta: pregnancy characteristics, maternal, and neonatal outcomes. American Journal of Obstetrics & Gynecology MFM, 2021, 3, 100362.	1.3	11
13	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	0.6	4
14	Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genetics in Medicine, 2020, 22, 490-499.	1.1	136
15	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
16	Fetal cardiac rhabdomyomas treated with maternal sirolimus. Prenatal Diagnosis, 2020, 40, 358-364.	1.1	17
17	Clinical Presentation of Coronavirus Disease 2019 (COVID-19) in Pregnant and Recently Pregnant People. Obstetrics and Gynecology, 2020, 136, 1117-1125.	1.2	70
18	Biallelic mutations in LAMA5 disrupts a skeletal noncanonical focal adhesion pathway and produces a distinct bent bone dysplasia. EBioMedicine, 2020, 62, 103075.	2.7	7

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19	Improvement in ventriculomegaly following cervicomedullary decompressive surgery in children with achondroplasia and foramen magnum stenosis. American Journal of Medical Genetics, Part A, 2020, 182, 1896-1905.	0.7	5
20	Gender representation of speakers at the Society for Maternal-Fetal Medicine postgraduate courses: a 20-year review. American Journal of Obstetrics & Gynecology MFM, 2020, 2, 100131.	1.3	1
21	Malocclusion traits and oral healthâ€“related quality of life in children with osteogenesis imperfecta. Journal of the American Dental Association, 2020, 151, 480-490.e2.	0.7	9
22	Mutations in GRK2 cause Jeune syndrome by impairing Hedgehog and canonical Wnt signaling. EMBO Molecular Medicine, 2020, 12, e11739.	3.3	16
23	4-PBA Treatment Improves Bone Phenotypes in the Aga2 Mouse Model of Osteogenesis Imperfecta. Journal of Bone and Mineral Research, 2020, 37, 675-686.	3.1	14
24	Dominantâ€•negativeSOX9mutations in campomelic dysplasia. Human Mutation, 2019, 40, 2344-2352.	1.1	20
25	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	0.8	6
26	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	0.7	431
27	The Î±2 chain of type IX collagen is essential for type IX collagen biosynthesis. American Journal of Medical Genetics, Part A, 2019, 179, 1672-1677.	0.7	1
28	259: Postpartum emergency department usage among women with psychiatric illness. American Journal of Obstetrics and Gynecology, 2019, 220, S187.	0.7	0
29	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
30	Fibroblast growth factor receptor influences primary cilium length through an interaction with intestinal cell kinase. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 4316-4325.	3.3	29
31	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
32	Firstâ€•Trimester Abdominal Circumference (Versus Crown Rump Length) Improves Precision in Interâ€•and Intraobserver Variability. Journal of Ultrasound in Medicine, 2019, 38, 2161-2167.	0.8	1
33	Oro-dental and cranio-facial characteristics of osteogenesis imperfecta type V. European Journal of Medical Genetics, 2019, 62, 103606.	0.7	11
34	A new biometric: In utero growth curves for metacarpal and phalangeal lengths reveal an embryonic patterning ratio. Prenatal Diagnosis, 2019, 39, 200-208.	1.1	1
35	NRP1 haploinsufficiency predisposes to the development of Tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2018, 176, 649-656.	0.7	4
36	Regulation of ciliary function by fibroblast growth factor signaling identifies FGFR3-related disorders achondroplasia and thanatophoric dysplasia as ciliopathies. Human Molecular Genetics, 2018, 27, 1093-1105.	1.4	33

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37	Expanding the genetic architecture and phenotypic spectrum in the skeletal ciliopathies. Human Mutation, 2018, 39, 152-166.	1.1	92
38	Proteomic analyses of signalling complexes associated with receptor tyrosine kinase identify novel members of fibroblast growth factor receptor 3 interactome. Cellular Signalling, 2018, 42, 144-154.	1.7	14
39	Atelosteogenesis Disorders. , 2018, , 254-256.e1.		0
40	Campomelic Dysplasia. , 2018, , 257-259.e1.		0
41	Short Rib Thoracic Dysplasia With or Without Polydactyly. , 2018, , 280-283.e1.		0
42	Spondyloepiphyseal Dysplasia Congenita. , 2018, , 283-285.e1.		0
43	FGFR3 Disorders. , 2018, , 264-267.e1.		1
44	The inositol phosphatase SHIP2 enables sustained ERK activation downstream of FGF receptors by recruiting Src kinases. Science Signaling, 2018, 11, .	1.6	14
45	The PTH/PTHrP-SIK3 pathway affects skeletogenesis through altered mTOR signaling. Science Translational Medicine, 2018, 10, .	5.8	38
46	DTDST Dysplasia (Including AOII and Achondrogenesis IB). , 2018, , 261-264.e1.		0
47	Chondrodysplasia Punctata. , 2018, , 259-261.e1.		0
48	Acrofacial Dysostosis. , 2018, , 288-291.e1.		0
49	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGFÎ² signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	1.6	29
50	Fkbp10 Deletion in Osteoblasts Leads to Qualitative Defects in Bone. Journal of Bone and Mineral Research, 2017, 32, 1354-1367.	3.1	16
51	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.	3.1	50
52	Mutations in IFT-A satellite core component genes IFT43 and IFT121 produce short rib polydactyly syndrome with distinctive campomelia. Cilia, 2017, 6, 7.	1.8	26
53	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	0.7	24
54	Mutations in <scp>DYNC2H1</scp>, the cytoplasmic dynein 2, heavy chain 1 motor protein gene, cause shortâ€rib polydactyly type I, Saldinoâ€“Noonan type. Clinical Genetics, 2017, 92, 158-165.	1.0	21

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55	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	18.1	481
56	<scp>MED</scp> resulting from recessively inherited mutations in the gene encoding calciumâ€activated nucleotidase <scp>CANT1</scp>. American Journal of Medical Genetics, Part A, 2017, 173, 2415-2421.	0.7	21
57	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€. American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
58	Genes uniquely expressed in human growth plate chondrocytes uncover a distinct regulatory network. BMC Genomics, 2017, 18, 983.	1.2	17
59	Heritable Diseases of Connective Tissue. , 2017, , 1797-1815.		0
60	Clinical and radiographic delineation of Bent Bone Dysplasiaâ€FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angelâ€shaped Phalanges. American Journal of Medical Genetics, Part A, 2016, 170, 2652-2661.	0.7	6
61	The ciliopathy-associated CPLANE proteins direct basal body recruitment of intraflagellar transport machinery. Nature Genetics, 2016, 48, 648-656.	9.4	119
62	An inactivating mutation in intestinal cell kinase,ICK, impairs hedgehog signalling and causes short rib-polydactyly syndrome. Human Molecular Genetics, 2016, 25, 3998-4011.	1.4	44
63	IFT52mutations destabilize anterograde complex assembly, disrupt ciliogenesis and result in short rib polydactyly syndrome. Human Molecular Genetics, 2016, 25, 4012-4020.	1.4	44
64	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	2.6	52
65	Somatic mosaicism for a lethal TRPV4 mutation results in nonâ€lethal metatropic dysplasia. American Journal of Medical Genetics, Part A, 2016, 170, 3298-3302.	0.7	8
66	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.	1.6	44
67	769: Clinical accuracy of abnormal cell-free fetal DNA (cfDNA) results for the sex chromosomes. American Journal of Obstetrics and Gynecology, 2016, 214, S402-S403.	0.7	2
68	618: Clinical accuracy of abnormal autosomal cell-free fetal DNA (cfDNA) screening. American Journal of Obstetrics and Gynecology, 2016, 214, S330.	0.7	1
69	TGFÎ² and BMP Dependent Cell Fate Changes Due to Loss of Filamin B Produces Disc Degeneration and Progressive Vertebral Fusions. PLoS Genetics, 2016, 12, e1005936.	1.5	47
70	Altered mRNA Splicing, Chondrocyte Gene Expression and Abnormal Skeletal Development due to SF3B4 Mutations in Rodriguez Acrofacial Dysostosis. PLoS Genetics, 2016, 12, e1006307.	1.5	48
71	A second locus for schneckenbecken dysplasia identified by a mutation in the gene encoding inositol polyphosphate phosphataseâ€like 1 (INPPL1). American Journal of Medical Genetics, Part A, 2015, 167, 2470-2473.	0.7	9
72	Skeletal Dysplasias. Clinics in Perinatology, 2015, 42, 301-319.	0.8	98

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73	Mutations in DYNC2LI1 disrupt cilia function and cause short rib polydactyly syndrome. Nature Communications, 2015, 6, 7092.	5.8	79
74	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
75	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	5.8	51
76	Detection of Y chromosome material in a 46,XX male with SRY translocation: novel application of cell-free fetal DNA testing. Prenatal Diagnosis, 2015, 35, 823-825.	1.1	2
77	HSP47 and FKBP65 cooperate in the synthesis of type I procollagen. Human Molecular Genetics, 2015, 24, 1918-1928.	1.4	50
78	Osteogenesis Imperfecta and Pregnancy. , 2014, , 243-251.		2
79	FKBP10 (FKBP65 Protein), Osteogenesis Imperfecta and Bruck Syndrome. , 2014, , 151-157.		0
80	Patient-Derived Skeletal Dysplasia Induced Pluripotent Stem Cells Display Abnormal Chondrogenic Marker Expression and Regulation byBMP2andTGFÎ²1. Stem Cells and Development, 2014, 23, 1464-1478.	1.1	44
81	Prenatal and postnatal findings in serpentine fibula polycystic kidney syndrome and a review of the NOTCH2 spectrum disorders. American Journal of Medical Genetics, Part A, 2014, 164, 2490-2495.	0.7	6
82	Opsismodysplasia resulting from an insertion mutation in the SH2 domain, which destabilizes INPPL1. American Journal of Medical Genetics, Part A, 2014, 164, 2407-2411.	0.7	10
83	Bent bone dysplasia syndrome reveals nucleolar activity for FGFR2 in ribosomal DNA transcription. Human Molecular Genetics, 2014, 23, 5659-5671.	1.4	36
84	Connective tissue alterations in Fkbp10â^'/â^' mice. Human Molecular Genetics, 2014, 23, 4822-4831.	1.4	54
85	Dynamic cervicomedullary cord compression and alterations in cerebrospinal fluid dynamics in children with achondroplasia: review of an 11-year surgical case series. Journal of Neurosurgery: Pediatrics, 2014, 14, 238-244.	0.8	30
86	Routine measurement of amniotic fluid alpha-fetoprotein and acetylcholinesterase: the need for a reevaluation. American Journal of Obstetrics and Gynecology, 2014, 211, 139.e1-139.e6.	0.7	28
87	The Dysostoses. , 2013, , 1-22.		1
88	WDR34 Mutations that Cause Short-Rib Polydactyly Syndrome Type III/Severe Asphyxiating Thoracic Dysplasia Reveal a Role for the NF-Î°B Pathway in Cilia. American Journal of Human Genetics, 2013, 93, 926-931.	2.6	79
89	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 137-143.	2.6	53
90	WNT1 Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	13.9	308

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91	Heritable Diseases of Connective Tissue. , 2013, , 1719-1739.e3.		2
92	Bent Bone Dysplasia Syndrome defines a nuclear role for FGFR2 in skeletal development. FASEB Journal, 2013, 27, 319.2.	0.2	0
93	A newly recognized syndrome with characteristic facial features, skeletal dysplasia, and developmental delay. American Journal of Medical Genetics, Part A, 2012, 158A, 1815-1822.	0.7	9
94	Bent Bone Dysplasia-FGFR2 type, a Distinct Skeletal Disorder, Has Deficient Canonical FGF Signaling. American Journal of Human Genetics, 2012, 90, 550-557.	2.6	74
95	Exome Sequencing Identifies PDE4D Mutations in Acrodysostosis. American Journal of Human Genetics, 2012, 90, 746-751.	2.6	128
96	Disease-associated mutations in the actin-binding domain of filamin B cause cytoplasmic focal accumulations correlating with disease severity. Human Mutation, 2012, 33, 665-673.	1.1	31
97	Dominant and recessive forms of fibrochondrogenesis resulting from mutations at a second locus, COL11A2. American Journal of Medical Genetics, Part A, 2012, 158A, 309-314.	0.7	16
98	The importance of conventional radiography in the mutational analysis of skeletal dysplasias (the) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.1	15
99	A novel skeletal disorder defines an intracellular role for FGFR2 during development. FASEB Journal, 2012, 26, 457.7.	0.2	0
100	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	1.5	45
101	Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	2.6	199
102	The skeleton and musculature on foetal MRI. Insights Into Imaging, 2011, 2, 309-318.	1.6	21
103	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	0.7	573
104	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	3.1	149
105	RefilinB (FAM101B) targets FilaminA to organize perinuclear actin networks and regulates nuclear shape. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 11464-11469.	3.3	78
106	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 551-559.	2.6	278
107	Response to Shaheen etÂal.. American Journal of Human Genetics, 2010, 87, 308.	2.6	2
108	BMPER Mutation in Diaphanospondylodysostosis Identified by Ancestral Autozygosity Mapping and Targeted High-Throughput Sequencing. American Journal of Human Genetics, 2010, 87, 532-537.	2.6	35

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109	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 572-573.	2.6	13
110	Fibrochondrogenesis Results from Mutations in the COL11A1 Type XI Collagen Gene. American Journal of Human Genetics, 2010, 87, 708-712.	2.6	69
111	Postnatal growth retardation, facial dysmorphism, spondylocarpal synostosis, cardiac defect, and inner ear malformation (cardiospondylocarpofacial syndrome?)â€"A distinct syndrome?. American Journal of Medical Genetics, Part A, 2010, 152A, 539-546.	0.7	8
112	Dominant TRPV4 mutations in nonlethal and lethal metatropic dysplasia. American Journal of Medical Genetics, Part A, 2010, 152A, 1169-1177.	0.7	93
113	Fetal alcohol syndrome: a phenocopy of spondylocarpotarsal synostosis syndrome?. Clinical Dysmorphology, 2010, 19, 175-180.	0.1	1
114	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	13.9	122
115	The skeletal dysplasias. Genetics in Medicine, 2010, 12, 327-341.	1.1	193
116	Generalized Connective Tissue Disease in Crtap-/- Mouse. PLoS ONE, 2010, 5, e10560.	1.1	52
117	Guidelines for the prenatal diagnosis of fetal skeletal dysplasias. Genetics in Medicine, 2009, 11, 127-133.	1.1	169
118	The Erlenmeyer flask bone deformity in the skeletal dysplasias. American Journal of Medical Genetics, Part A, 2009, 149A, 1334-1345.	0.7	55
119	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, 72-79.	2.6	120
120	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, 307-315.	2.6	173
121	Ciliary Abnormalities Due to Defects in the Retrograde Transport Protein DYNC2H1 in Short-Rib Polydactyly Syndrome. American Journal of Human Genetics, 2009, 84, 542-549.	2.6	149
122	Heritable Diseases of Connective Tissue. , 2009, , 1635-1655.		1
123	CRTAP and LEPRE1 mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	1.1	196
124	Expanded clinical spectrum of spondylocarpotarsal synostosis syndrome and possible manifestation in a heterozygous father. American Journal of Medical Genetics, Part A, 2008, 146A, 779-783.	0.7	19
125	Evaluation of prenatalâ€onset osteochondrodysplasias by ultrasonography: A retrospective and prospective analysis. American Journal of Medical Genetics, Part A, 2008, 146A, 1917-1924.	0.7	77
126	Spondylocarpotarsal synostosis: Longâ€term followâ€up of a case due to FLNB mutations. American Journal of Medical Genetics, Part A, 2008, 146A, 1230-1233.	0.7	6

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127	ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF-Î² bioavailability regulation. Nature Genetics, 2008, 40, 1119-1123.	9.4	211
128	Sprouty 2 disturbs FGFR3 degradation in thanatophoric dysplasia type II: A severe form of human achondroplasia. Cellular Signalling, 2008, 20, 1471-1477.	1.7	33
129	Fibroblast Growth Factors 1, 2, 17, and 19 Are the Predominant FGF Ligands Expressed in Human Fetal Growth Plate Cartilage. Pediatric Research, 2007, 61, 267-272.	1.1	55
130	Disruption of the Flnb gene in mice phenocopies the human disease spondylocarpotarsal synostosis syndrome. Human Molecular Genetics, 2007, 17, 631-641.	1.4	51
131	Type XXVII collagen at the transition of cartilage to bone during skeletogenesis. Bone, 2007, 41, 535-542.	1.4	67
132	Angulated femurs and the skeletal dysplasias: Experience of the International Skeletal Dysplasia Registry (1988â€“2006). American Journal of Medical Genetics, Part A, 2007, 143A, 1159-1168.	0.7	33
133	Consortium for osteogenesis imperfecta mutations in the helical domain of type I collagen: regions rich in lethal mutations align with collagen binding sites for integrins and proteoglycans. Human Mutation, 2007, 28, 209-221.	1.1	620
134	Cartilage-selective genes identified in genome-scale analysis of non-cartilage and cartilage gene expression. BMC Genomics, 2007, 8, 165.	1.2	19
135	The Skeletal Dysplasias. Annals of the New York Academy of Sciences, 2007, 1117, 302-309.	1.8	65
136	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	1.5	102
137	GDF5 Is a Second Locus for Multiple-Synostosis Syndrome. American Journal of Human Genetics, 2006, 78, 708-712.	2.6	83
138	Obstetrics and obstetrical anesthesia issues in women with DWARFISM. American Journal of Obstetrics and Gynecology, 2006, 195, S185.	0.7	4
139	Terminal phalangeal accessory ossification center of the thumb: an additional radiographic finding in Larsen syndrome. Pediatric Radiology, 2006, 36, 970-973.	1.1	5
140	Mutations in two regions ofFLNBresult in atelosteogenesis I and III. Human Mutation, 2006, 27, 705-710.	1.1	66
141	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	0.7	67
142	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity (Am J Med Genet 140A:) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.7	2
143	Dominance of SOX9 function over RUNX2 during skeletogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 19004-19009.	3.3	325
144	MED, COMP, multilayered and NEIN: an overview of multiple epiphyseal dysplasia. Pediatric Radiology, 2005, 35, 116-123.	1.1	46

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145	Dysregulation of Chondrogenesis in Human Cleidocranial Dysplasia. American Journal of Human Genetics, 2005, 77, 305-312.	2.6	45
146	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	9.4	252
147	A transcriptional profile of human fetal cartilage. Matrix Biology, 2004, 23, 299-307.	1.5	19
148	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.	0.5	10
149	Radiographic findings and Gs-alpha bioactivity studies and mutation screening in acrodysostosis indicate a different etiology from pseudohypoparathyroidism. Pediatric Radiology, 2001, 31, 2-9.	1.1	33
150	Double heterozygosity for pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. American Journal of Medical Genetics Part A, 2001, 104, 140-146.	2.4	42
151	Widely distributed mutations in the COL2A1 gene produce achondrogenesis type II/hypochondrogenesis. , 2000, 92, 95-100.		60
152	Exclusion of the Ellisâ€“van Creveld region on chromosomeÂ4p16 in some families with asphyxiating thoracic dystrophy and short-rib polydactyly syndromes. European Journal of Human Genetics, 2000, 8, 645-648.	1.4	13
153	Heterozygous mutations in the gene encoding noggin affect human joint morphogenesis. Nature Genetics, 1999, 21, 302-304.	9.4	329
154	Human Ehlers-Danlos Syndrome Type VII C and Bovine Dermatosparaxis Are Caused by Mutations in the Procollagen I N-Proteinase Gene. American Journal of Human Genetics, 1999, 65, 308-317.	2.6	348
155	Molecular, radiologic, and histopathologic correlations in thanatophoric dysplasia. , 1998, 78, 274-281.		127
156	Effect of hemodialysis on uterine and umbilical artery Doppler flow velocity waveforms. American Journal of Obstetrics and Gynecology, 1994, 170, 1386-1388.	0.7	8
157	Genetic aspects of ovarian cancer. Current Opinion in Obstetrics and Gynecology, 1994, 6, 105.	0.9	4
158	COFFEE AND PANCREATIC CANCER: AN ANALYSIS OF INTERNATIONAL MORTALITY DATA1. American Journal of Epidemiology, 1983, 118, 630-640.	1.6	31
159	Differential diagnosis III: osteogenesis imperfecta. , 0, , 254-267.		2