

# Nosology and classification of genetic skeletal disorders

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Spondyloepimetaphyseal dysplasia with joint laxity (Beighton type): A unique South African disorder. South African Medical Journal, 2016, 106, 54.	0.2	6
2	Osteogenesis imperfecta in southern Africa: Peter Beighton's legacy. South African Medical Journal, 2016, 106, 13.	0.2	0
3	Genetics of osteoporosis: searching for candidate genes for bone fragility. Archives of Endocrinology and Metabolism, 2016, 60, 391-401.	0.3	37
4	Growth Hormone Axis in Skeletal Dysplasias. , 0, , .		0
5	A homozygous nonsense variant in <i>IFT52</i> is associated with a human skeletal ciliopathy. Clinical Genetics, 2016, 90, 536-539.	1.0	68
6	Characterization of a Relatively Malignant Form of Osteopetrosis Caused by a Novel Mutation in the <i>PLEKHM1</i> Gene. Journal of Bone and Mineral Research, 2016, 31, 1979-1987.	3.1	26
7	Skeletal Dysplasias That Cause Thoracic Insufficiency in Neonates. Medicine (United States), 2016, 95, e3298.	0.4	6
8	StÃ¼ve-Wiedemann Syndrome: Update on Clinical and Genetic Aspects. Molecular Syndromology, 2016, 7, 12-18.	0.3	18
9	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	0.7	31
10	Metatropic dysplasia is associated with increased fracture risk. American Journal of Medical Genetics, Part A, 2016, 170, 1373-1376.	0.7	2
11	Desbuquois dysplasia type II in a patient with a homozygous mutation in <i>XYLT1</i> and new unusual findings. American Journal of Medical Genetics, Part A, 2016, 170, 3043-3047.	0.7	14
12	Skeletal dysplasias: New medical treatments. Anales De PediatrÃa (English Edition), 2016, 85, 1-3.	0.1	0
14	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	9.4	125
15	Spondyloepiphyseal Dysplasia Congenita in a painting of Vicente LÃ³pez y PortaÃ±a (1825). Journal of Endocrinological Investigation, 2016, 39, 717-717.	1.8	1
16	Osteoporosis and Bone Mass Disorders: From Gene Pathways to Treatments. Trends in Endocrinology and Metabolism, 2016, 27, 262-281.	3.1	108
17	Osteogenesis imperfecta in children and adolescents" new developments in diagnosis and treatment. Osteoporosis International, 2016, 27, 3427-3437.	1.3	150
18	An inactivating mutation in intestinal cell kinase, <i>ICK</i> , impairs hedgehog signalling and causes short rib-polydactyly syndrome. Human Molecular Genetics, 2016, 25, 3998-4011.	1.4	44
19	From pseudohypoparathyroidism to inactivating PTH/PTHrP signalling disorder (iPPSD), a novel classification proposed by the EuroPHP network. European Journal of Endocrinology, 2016, 175, P1-P17.	1.9	117

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20	DNA sequence analysis in 598 individuals with a clinical diagnosis of osteogenesis imperfecta: diagnostic yield and mutation spectrum. <i>Osteoporosis International</i> , 2016, 27, 3607-3613.	1.3	119
21	Diagnostic conundrums in antenatal presentation of a skeletal dysplasia with description of a heterozygous C-terminal propeptide mutation in <i>COL1A1</i> associated with a severe presentation of osteogenesis imperfecta. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3303-3307.	0.7	2
22	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. <i>American Journal of Human Genetics</i> , 2016, 99, 392-406.	2.6	52
23	Skeletal dysplasia. A guide to the orthopaedic surgeon. <i>Orthopaedics and Trauma</i> , 2016, 30, 500-517.	0.2	0
24	Destabilization of the IFT-B cilia core complex due to mutations in IFT81 causes a Spectrum of Short-Rib Polydactyly Syndrome. <i>Scientific Reports</i> , 2016, 6, 34232.	1.6	44
25	Rehabilitation for Patients with Skeletal Dysplasias. <i>The Japanese Journal of Rehabilitation Medicine</i> , 2016, 53, 374-378.	0.0	0
26	A novel type II collagen gene mutation in a family with spondyloepiphyseal dysplasia and extensive intrafamilial phenotypic diversity. <i>Human Genome Variation</i> , 2016, 3, 16007.	0.4	3
27	Molecular analysis of the CTSK gene in a cohort of 33 Brazilian families with pycnodysostosis from a cluster in a Brazilian Northeast region. <i>European Journal of Medical Research</i> , 2016, 21, 33.	0.9	18
28	Skeleton Genetics: a comprehensive database for genes and mutations related to genetic skeletal disorders. <i>Database: the Journal of Biological Databases and Curation</i> , 2016, 2016, baw127.	1.4	12
29	Challenges in the Management of Short Stature. <i>Hormone Research in Paediatrics</i> , 2016, 85, 2-10.	0.8	47
30	Cortical-Bone Fragility – Insights from sFRP4 Deficiency in Pyle's Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2553-2562.	13.9	119
31	Brief Report: Peripheral Osteolysis in Adults Linked to <i>ASAH1</i> (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. <i>Arthritis and Rheumatology</i> , 2016, 68, 2323-2327.	2.9	17
32	Mechanism of pancreatic and liver malformations in human fetuses with short-rib polydactyly syndrome. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2016, 106, 549-562.	1.6	1
33	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. <i>European Journal of Human Genetics</i> , 2016, 24, 1206-1210.	1.4	16
34	MECHANISMS IN ENDOCRINOLOGY: Novel genetic causes of short stature. <i>European Journal of Endocrinology</i> , 2016, 174, R145-R173.	1.9	134
35	BMP signalling in skeletal development, disease and repair. <i>Nature Reviews Endocrinology</i> , 2016, 12, 203-221.	4.3	607
36	Two novel mutations in TMEM38B result in rare autosomal recessive osteogenesis imperfecta. <i>Journal of Human Genetics</i> , 2016, 61, 539-545.	1.1	30
37	Genetic control of bone mass. <i>Molecular and Cellular Endocrinology</i> , 2016, 432, 3-13.	1.6	59

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38	Exome sequencing revealed a novel splice site variant in the <i>ALX1</i> gene underlying frontonasal dysplasia. <i>Clinical Genetics</i> , 2017, 91, 494-498.	1.0	13
39	Spondyloepimetaphyseal dysplasia with neurodegeneration associated with <i>AIFM1</i> mutation – a novel phenotype of the mitochondrial disease. <i>Clinical Genetics</i> , 2017, 91, 30-37.	1.0	38
40	Skeletal dysplasia. <i>Surgery</i> , 2017, 35, 52-61.	0.1	0
41	Broadening the phenotypic spectrum of <i>POP1</i> skeletal dysplasias: identification of <i>POP1</i> mutations in a mild and severe skeletal dysplasia. <i>Clinical Genetics</i> , 2017, 92, 91-98.	1.0	19
42	A framework for the classification of joint hypermobility and related conditions. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 148-157.	0.7	356
43	<i>EXTL3</i> mutations cause skeletal dysplasia, immune deficiency, and developmental delay. <i>Journal of Experimental Medicine</i> , 2017, 214, 623-637.	4.2	76
44	Axial spondylometaphyseal dysplasia is also caused by <i>NEK1</i> mutations. <i>Journal of Human Genetics</i> , 2017, 62, 503-506.	1.1	25
45	Echocardiographic phenotype in osteogenesis imperfecta varies with disease severity. <i>Heart</i> , 2017, 103, 443-448.	1.2	13
46	Three-dimensional ultrasound imaging of the fetal skull and face. <i>Ultrasound in Obstetrics and Gynecology</i> , 2017, 50, 7-16.	0.9	29
47	Bone robusticity in two distinct skeletal dysplasias diverges from established patterns. <i>Journal of Orthopaedic Research</i> , 2017, 35, 2392-2396.	1.2	6
48	Mutations in IFT-A satellite core component genes <i>IFT43</i> and <i>IFT121</i> produce short rib polydactyly syndrome with distinctive campomelia. <i>Cilia</i> , 2017, 6, 7.	1.8	26
49	Metatropic dysplasia in third trimester of pregnancy and a novel causative variant in the <i>TRPV4</i> gene. <i>European Journal of Medical Genetics</i> , 2017, 60, 365-368.	0.7	2
50	Bone dysplasia. <i>Annales D'Endocrinologie</i> , 2017, 78, 114-122.	0.6	2
51	Ultra-Low-Dose Fetal CT With Model-Based Iterative Reconstruction: A Prospective Pilot Study. <i>American Journal of Roentgenology</i> , 2017, 208, 1365-1372.	1.0	20
52	Next-Generation Sequencing Based Testing for Disorders of the Skeleton. , 2017, , 113-121.		0
53	Severe generalised chondrodysplasia in miniature cattle breeds. <i>New Zealand Veterinary Journal</i> , 2017, 65, 282-283.	0.4	4
54	Airway stenting in a child with spondyloepiphyseal dysplasia congenita: 13-Year survival. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2017, 99, 13-16.	0.4	1
55	Grebe dysplasia – prenatal diagnosis based on rendered 3-D ultrasound images of fetal limbs. <i>Pediatric Radiology</i> , 2017, 47, 108-112.	1.1	2

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56	Perinatal imaging findings and molecular genetic analysis of Athanaphoric dysplasia type 1 in a fetus with a c.2419T>G (p.Ter807Gly) (X807G) mutation in FGFR3. Taiwanese Journal of Obstetrics and Gynecology, 2017, 56, 87-92.	0.5	3
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58	Follow-Up Study on Fetal CT Radiation Dose in Japan: Validating the Decrease in Radiation Dose. American Journal of Roentgenology, 2017, 208, 862-867.	1.0	3
59	Ossification center of the humeral shaft in the human fetus: a CT, digital, and statistical study. Surgical and Radiologic Anatomy, 2017, 39, 1107-1116.	0.6	8
60	The potential of induced pluripotent stem cells as a tool to study skeletal dysplasias and cartilage-related pathologic conditions. Osteoarthritis and Cartilage, 2017, 25, 616-624.	0.6	17
61	Neuroimaging Findings in Pediatric Genetic Skeletal Disorders: A Review. Journal of Neuroimaging, 2017, 27, 162-209.	1.0	6
62	Mutations in <i>DYNC2H1</i> , the cytoplasmic dynein 2, heavy chain 1 motor protein gene, cause short-rib polydactyly type I, Saldino-Noonan type. Clinical Genetics, 2017, 92, 158-165.	1.0	21
63	Three cases of Japanese acromicric/geleophysic dysplasia with FBN1 mutations: a comparison of clinical and radiological features. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 117-121.	0.4	8
64	Heritable Skeletal Disorders Arising from Defects in Processing and Transport of Type I Procollagen from the ER: Perspectives on Possible Therapeutic Approaches. Handbook of Experimental Pharmacology, 2017, 245, 191-225.	0.9	3
65	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	2.6	90
66	Osteogenesis imperfecta. Nature Reviews Disease Primers, 2017, 3, 17052.	18.1	481
67	Long-Term Bisphosphonate Therapy in Osteogenesis Imperfecta. Current Osteoporosis Reports, 2017, 15, 412-418.	1.5	50
68	ARQ 087 inhibits FGFR signaling and rescues aberrant cell proliferation and differentiation in experimental models of craniosynostoses and chondrodysplasias caused by activating mutations in FGFR1, FGFR2 and FGFR3. Bone, 2017, 105, 57-66.	1.4	17
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70	Genetic screening confirms heterozygous mutations in ACAN as a major cause of idiopathic short stature. Scientific Reports, 2017, 7, 12225.	1.6	53
71	Osteogenesis imperfecta: diagnosis and treatment. Current Opinion in Endocrinology, Diabetes and Obesity, 2017, 24, 381-388.	1.2	69
72	Additional report on Moreno-Nishimura-Schmidt overgrowth syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2834-2837.	0.7	1
73	Genetic disorders of bone - An historical perspective. Bone, 2017, 102, 1-4.	1.4	5

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74	Expression of RMRP RNA is regulated in chondrocyte hypertrophy and determines chondrogenic differentiation. <i>Scientific Reports</i> , 2017, 7, 6440.	1.6	43
75	Best practices in perioperative management of patients with skeletal dysplasias. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2584-2595.	0.7	31
76	A case report of pycnodysostosis with atypical femur fracture diagnosed by next-generation sequencing of candidate genes. <i>Medicine (United States)</i> , 2017, 96, e6367.	0.4	20
77	Skeletal Dysplasias: What Every Bone Health Clinician Needs to Know. <i>Current Osteoporosis Reports</i> , 2017, 15, 419-424.	1.5	8
78	Fetal Skeletal Lethal Dysplasia: Case Report. <i>Revista Brasileira De Ginecologia E Obstetricia</i> , 2017, 39, 576-582.	0.3	0
79	A Novel de novo FZD2 Mutation in a Patient with Autosomal Dominant Omodysplasia. <i>Molecular Syndromology</i> , 2017, 8, 318-324.	0.3	16
80	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. <i>Scientific Reports</i> , 2017, 7, 15585.	1.6	21
81	Recent Discoveries in Monogenic Disorders of Childhood Bone Fragility. <i>Current Osteoporosis Reports</i> , 2017, 15, 303-310.	1.5	16
82	Delineation of Ehlers-Danlos syndrome phenotype due to the c.934C>T, p.(Arg312Cys) mutation in <i>COL1A1</i> : Report on a three-generation family without cardiovascular events, and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 524-530.	0.7	35
83	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. <i>Journal of Human Genetics</i> , 2017, 62, 437-441.	1.1	33
84	Chondroitin Sulfate N-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. <i>Human Mutation</i> , 2017, 38, 34-38.	1.1	22
85	Transcriptional control of chondrocyte specification and differentiation. <i>Seminars in Cell and Developmental Biology</i> , 2017, 62, 34-49.	2.3	142
86	Genomic approaches to diagnose rare bone disorders. <i>Bone</i> , 2017, 102, 5-14.	1.4	15
87	The Bone in Genetic and Metabolic Diseases: A Practical Approach. , 2017, , 371-380.		0
88	INPPL1 gene mutations in opsismodysplasia. <i>Journal of Human Genetics</i> , 2017, 62, 135-140.	1.1	13
89	Mutation analysis of <i>FGFR1</i> in 11 Japanese patients with syndromic craniosynostoses. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 157-162.	0.7	7
90	Signaling pathways regulating cartilage growth plate formation and activity. <i>Seminars in Cell and Developmental Biology</i> , 2017, 62, 3-15.	2.3	53
91	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 733-739.	0.7	8

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92	Novel and recurrent XYLT1 mutations in two Turkish families with Desbuquois dysplasia, type 2. <i>Journal of Human Genetics</i> , 2017, 62, 447-451.	1.1	24
93	Craniofacial manifestations in osteogenesis imperfecta type III in South Africa. <i>BDJ Open</i> , 2017, 3, 17021.	0.8	8
94	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with <i>B3GALT6</i> Mutations. <i>Molecular Syndromology</i> , 2017, 8, 303-307.	0.3	7
95	Skeletal Dysplasias: Growing Therapy for Growing Bones. <i>Frontiers in Pharmacology</i> , 2017, 8, 79.	1.6	8
96	Severe Congenital Neutropenias and Other Rare Inherited Disorders With Marrow Failure. , 2017, , 241-253.		1
97	Clinical and radiographic features of pycnodysostosis: A case report. <i>Journal of Clinical and Experimental Dentistry</i> , 2017, 9, e1276-e1281.	0.5	12
98	Genes uniquely expressed in human growth plate chondrocytes uncover a distinct regulatory network. <i>BMC Genomics</i> , 2017, 18, 983.	1.2	17
100	Symmetrical brachydactyly in a dog. <i>Veterinary and Comparative Orthopaedics and Traumatology</i> , 2017, 30, 306-309.	0.2	1
101	Early Osteoarthritis and Double-Layered Patella in a Patient With Multiple Epiphyseal Dysplasia. <i>Archives of Rheumatology</i> , 2017, 32, 260-263.	0.3	1
102	Screening Gene Knockout Mice for Variation in Bone Mass: Analysis by $\mu$ CT and Histomorphometry. <i>Current Osteoporosis Reports</i> , 2018, 16, 77-94.	1.5	28
103	Translational studies provide insights for the etiology and treatment of cortical bone osteoporosis. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2018, 32, 329-340.	2.2	19
104	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. <i>Bone</i> , 2018, 110, 368-377.	1.4	38
105	Osteogenesis Imperfecta Type I: Recognition in Primary Care. <i>Journal for Nurse Practitioners</i> , 2018, 14, 470-476.	0.4	0
108	Expansion of the clinical spectrum of frontometaphyseal dysplasia 2 caused by the recurrent mutation p.Pro485Leu in MAP3K7. <i>European Journal of Medical Genetics</i> , 2018, 61, 612-615.	0.7	4
109	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018, 20, 1609-1616.	1.1	46
110	Somatic activating mutations in MAP2K1 cause melorheostosis. <i>Nature Communications</i> , 2018, 9, 1390.	5.8	56
111	Multicentric carpotarsal osteolysis syndrome: long-term follow-up of three patients. <i>Skeletal Radiology</i> , 2018, 47, 1015-1019.	1.2	10
112	Regulation of ciliary function by fibroblast growth factor signaling identifies FGFR3-related disorders achondroplasia and thanatophoric dysplasia as ciliopathies. <i>Human Molecular Genetics</i> , 2018, 27, 1093-1105.	1.4	33

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113	IHH Gene Mutations Causing Short Stature With Nonspecific Skeletal Abnormalities and Response to Growth Hormone Therapy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 604-614.	1.8	48
114	Targeted gene panel sequencing prenatally detects two novel mutations of <i>DYNC2H1</i> in a fetus with increased biparietal diameter and polyhydramnios. <i>Birth Defects Research</i> , 2018, 110, 364-371.	0.8	6
115	Severe rhizomelic shortening in a child with a complex duplication/deletion rearrangement of chromosome X. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 450-454.	0.7	3
116	A balanced reciprocal translocation t(10;15)(q22.3;q26.1) interrupting ACAN gene in a family with proportionate short stature. <i>Journal of Endocrinological Investigation</i> , 2018, 41, 929-936.	1.8	11
117	A novel homozygous variant in <i>BMPR1B</i> underlies acromesomelic dysplasia Hunter–Thompson type. <i>Annals of Human Genetics</i> , 2018, 82, 129-134.	0.3	13
118	Enzyme replacement therapy for mucopolysaccharidosis type IV (Morquio syndrome). <i>The Cochrane Library</i> , 2018, , .	1.5	1
119	Fetal Mesenchymal Stromal Cells: an Opportunity for Prenatal Cellular Therapy. <i>Current Stem Cell Reports</i> , 2018, 4, 61-68.	0.7	32
120	Dysosteosclerosis is also caused by TNFRSF11A mutation. <i>Journal of Human Genetics</i> , 2018, 63, 769-774.	1.1	21
121	What Is New in Prenatal Skeletal Dysplasias?. <i>American Journal of Roentgenology</i> , 2018, 210, 1022-1033.	1.0	18
122	Whole-genome sequencing of Atacama skeleton shows novel mutations linked with dysplasia. <i>Genome Research</i> , 2018, 28, 423-431.	2.4	19
123	Osteogenesis imperfecta and therapeutics. <i>Matrix Biology</i> , 2018, 71-72, 294-312.	1.5	75
124	Cartilage oligomeric matrix protein: COMPopathies and beyond. <i>Matrix Biology</i> , 2018, 71-72, 161-173.	1.5	131
125	Osteogenesis imperfecta – A clinical update. <i>Metabolism: Clinical and Experimental</i> , 2018, 80, 27-37.	1.5	103
126	Expanding the genetic architecture and phenotypic spectrum in the skeletal ciliopathies. <i>Human Mutation</i> , 2018, 39, 152-166.	1.1	92
127	Proteomic analyses of signalling complexes associated with receptor tyrosine kinase identify novel members of fibroblast growth factor receptor 3 interactome. <i>Cellular Signalling</i> , 2018, 42, 144-154.	1.7	14
128	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. <i>Genetics in Medicine</i> , 2018, 20, 630-638.	1.1	101
129	Outliers of bone metabolic diseases. <i>Metabolism: Clinical and Experimental</i> , 2018, 80, 1-4.	1.5	4
130	Novel spondyloepimetaphyseal dysplasia due to <i>UFSP2</i> gene mutation. <i>Clinical Genetics</i> , 2018, 93, 671-674.	1.0	26



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131	Genetics of clubfoot; recent progress and future perspectives. <i>European Journal of Medical Genetics</i> , 2018, 61, 107-113.	0.7	42
132	Skeletal dysplasias: an overview. <i>Paediatrics and Child Health (United Kingdom)</i> , 2018, 28, 84-92.	0.2	8
133	Preaxial polydactyly of the foot. <i>Monthly Notices of the Royal Astronomical Society: Letters</i> , 2018, 89, 113-118.	1.2	14
134	Genetic analysis of osteogenesis imperfecta in the <sc>P</sc>alestinian population: molecular screening of 49 affected families. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 15-26.	0.6	26
135	Mucopolysaccharidoses and Orthopedic Management (Focused also on Craniocervical Junction). <i>Journal of Child Science</i> , 2018, 08, e128-e137.	0.1	2
136	Children Born Small for Gestational Age: Differential Diagnosis, Molecular Genetic Evaluation, and Implications. <i>Endocrine Reviews</i> , 2018, 39, 851-894.	8.9	122
137	Dysmorphic Short Stature: Radiological Diagnosis of Trichorhinophalangeal Syndrome. <i>Case Reports in Pediatrics</i> , 2018, 2018, 1-5.	0.2	1
138	Short Rib Thoracic Dysplasia With or Without Polydactyly. , 2018, , 280-283.e1.		0
139	Clinical Diagnosis of X-Linked Spondyloepiphyseal Dysplasia Tarda and a Novel Missense Mutation in the Sedlin Gene (SEDL). <i>International Journal of Endocrinology</i> , 2018, 2018, 1-7.	0.6	0
140	Different Forms of ER Stress in Chondrocytes Result in Short Stature Disorders and Degenerative Cartilage Diseases: New Insights by Cartilage-Specific ERp57 Knockout Mice. <i>Oxidative Medicine and Cellular Longevity</i> , 2018, 2018, 1-14.	1.9	18
141	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. <i>American Journal of Human Genetics</i> , 2018, 103, 553-567.	2.6	58
142	Congenital limb deficiency in Japan: a cross-sectional nationwide survey on its epidemiology. <i>BMC Musculoskeletal Disorders</i> , 2018, 19, 262.	0.8	19
143	Confirmation of spondyloâ€piâ€metaphyseal dysplasia with joint laxity, <i>EXOC6B</i> type. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2934-2935.	0.7	5
144	Fetal Skeletal Disorders. <i>Ultraschall in Der Medizin</i> , 2018, 39, 610-634.	0.8	14
145	FGFR3 Disorders. , 2018, , 264-267.e1.		1
148	Best practice guidelines regarding prenatal evaluation and delivery of patients with skeletal dysplasia. <i>American Journal of Obstetrics and Gynecology</i> , 2018, 219, 545-562.	0.7	29
149	The inositol phosphatase SHIP2 enables sustained ERK activation downstream of FGF receptors by recruiting Src kinases. <i>Science Signaling</i> , 2018, 11, .	1.6	14
150	Screening of known disease genes in congenital scoliosis. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 966-974.	0.6	20

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152	Skeletal Dysplasias. , 2018, , 175-196.		1
153	Synergistic co-regulation and competition by a SOX9-GLI-FOXA phasic transcriptional network coordinate chondrocyte differentiation transitions. PLoS Genetics, 2018, 14, e1007346.	1.5	56
154	An Update on Osteomyelitis Treatment in a Pycnodysostosis Patient. Journal of Oral and Maxillofacial Surgery, 2018, 76, 2136.e1-2136.e10.	0.5	7
155	Biallelic B3GALT6 mutations cause spondylodysplastic Ehlersâ€“Danlos syndrome. Human Molecular Genetics, 2018, 27, 3475-3487.	1.4	34
156	Skeletal Dysplasias. , 2018, , 469-480.		1
157	Sclerosing Bone Disorders. , 2018, , 507-521.		1
158	Introduction to Genetics of Skeletal and Mineral Metabolic Diseases. , 2018, , 1-21.		2
159	Sclerosing bone dysplasias. Best Practice and Research in Clinical Endocrinology and Metabolism, 2018, 32, 707-723.	2.2	15
160	Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. Bone, 2018, 114, 125-136.	1.4	24
161	Further delineation of spondyloepimetaphyseal dysplasia Fadenâ€“Alkuraya type: A RSPRY1â€“associated spondyloâ€“epiâ€“metaphyseal dysplasia with conoâ€“brachydactyly and craniosynostosis. American Journal of Medical Genetics, Part A, 2018, 176, 2009-2016.	0.7	1
162	Clinical exome sequencing in France and Quebec: what are the challenges? What does the future hold?. Life Sciences, Society and Policy, 2018, 14, 17.	3.1	2
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