Sheila L Unger

List of Publications by Year
in descending order

142 papers	6,904 citations	57758 44 h-index	69250 77 g-index
148 all docs	148 docs citations	148 times ranked	9104 citing authors

#	Article	IF	Citations
1	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	1.2	573
2	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	1.2	453
3	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
4	Nosology and classification of genetic skeletal disorders: 2006 revision. American Journal of Medical Genetics, Part A, 2007, 143A, 1-18.	1.2	301
5	The Zinc Transporter SLC39A13/ZIP13 Is Required for Connective Tissue Development; Its Involvement in BMP/TGF-Î² Signaling Pathways. PLoS ONE, 2008, 3, e3642.	2.5	240
6	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.	6.2	199
7	Null Leukemia Inhibitory Factor Receptor (LIFR) Mutations in StÃ¼ve-Wiedemann/Schwartz-Jampel Type 2 Syndrome. American Journal of Human Genetics, 2004, 74, 298-305.	6.2	162
8	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	21.4	151
9	Pseudoachondroplasia and multiple epiphyseal dysplasia: New etiologic developments. American Journal of Medical Genetics Part A, 2001, 106, 244-250.	2.4	125
10	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
11	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	27.0	119
12	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	6.2	114
13	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	6.2	112
14	Mutations in the Heparan-Sulfate Proteoglycan Glypican 6 (GPC6) Impair Endochondral Ossification and Cause Recessive Omodysplasia. American Journal of Human Genetics, 2009, 84, 760-770.	6.2	106
15	Pseudoachondroplasia and multiple epiphyseal dysplasia: A 7â€year comprehensive analysis of the known disease genes identify novel and recurrent mutations and provides an accurate assessment of their relative contribution. Human Mutation, 2012, 33, 144-157.	2.5	104
16	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102
17	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. Genetics in Medicine, 2017, 19, 386-395.	2.4	94
18	Congenital Joint Dislocations Caused by Carbohydrate Sulfotransferase 3 Deficiency in Recessive Larsen Syndrome and Humero-Spinal Dysostosis. American Journal of Human Genetics, 2008, 82, 1368-1374.	6.2	92

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19	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	6.2	88
20	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, 217-229.	1.6	74
21	CDK10/cyclin M is a protein kinase that controls ETS2 degradation and is deficient in STAR syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 19525-19530.	7.1	73
22	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	1.6	71
23	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. Human Genetics, 2011, 129, 497-502.	3.8	68
24	Phenotypic features of carbohydrate sulfotransferase 3 (CHST3) deficiency in 24 patients: Congenital dislocations and vertebral changes as principal diagnostic features. American Journal of Medical Genetics, Part A, 2010, 152A, 2543-2549.	1.2	67
25	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	2.5	67
26	NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system, and retina. American Journal of Medical Genetics, Part A, 2015, 167, 2902-2912.	1.2	66
27	Mutations in the heat-shock protein A9 (HSPA9) gene cause the EVEN-PLUS syndrome of congenital malformations and skeletal dysplasia. Scientific Reports, 2015, 5, 17154.	3.3	65
28	Multiple epiphyseal dysplasia: radiographic abnormalities correlated with genotype. Pediatric Radiology, 2001, 31, 10-18.	2.0	61
29	Mutations in LONP1, a mitochondrial matrix protease, cause CODAS syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1501-1509.	1.2	61
30	TBX15 Mutations Cause Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature in Cousin Syndrome. American Journal of Human Genetics, 2008, 83, 649-655.	6.2	60
31	Spondyloenchondrodysplasia with spasticity, cerebral calcifications, and immune dysregulation: Clinical and radiographic delineation of a pleiotropic disorder. American Journal of Medical Genetics, Part A, 2006, 140A, 541-550.	1.2	58
32	Novel and recurrent TRPV4 mutations and their association with distinct phenotypes within the TRPV4 dysplasia family. Journal of Medical Genetics, 2010, 47, 704-709.	3.2	58
33	Evolutionary Comparison Provides Evidence for Pathogenicity of RMRP Mutations. PLoS Genetics, 2005, 1, e47.	3.5	57
34	Defective C-propeptides of the ProÎ±2(I) Chain of Type I Procollagen Impede Molecular Assembly and Result in Osteogenesis Imperfecta. Journal of Biological Chemistry, 2008, 283, 16061-16067.	3.4	57
35	Multiple epiphyseal dysplasia: clinical and radiographic features, differential diagnosis and molecular basis. Best Practice and Research in Clinical Rheumatology, 2008, 22, 19-32.	3.3	56
36	Spondyloâ€epiphyseal dysplasia, Maroteaux type (pseudoâ€Morquio syndrome type 2), and parastremmatic dysplasia are caused by TRPV4 mutations. American Journal of Medical Genetics, Part A, 2010, 152A, 1443-1449.	1.2	56

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37	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
38	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. Nature Communications, 2017, 8, 15861.	12.8	56
39	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. American Journal of Human Genetics, 2009, 85, 168-178.	6.2	54
40	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, 419-428.	6.2	53
41	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 137-143.	6.2	53
42	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
43	Current Care and Investigational Therapies in Achondroplasia. Current Osteoporosis Reports, 2017, 15, 53-60.	3.6	50
44	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. European Journal of Human Genetics, 2009, 17, 395-400.	2.8	48
45	Wholeâ€exome sequencing detects somatic mutations of IDH1 in metaphyseal chondromatosis with <scp>D</scp>â€2â€hydroxyglutaric aciduria (MCâ€HGA). American Journal of Medical Genetics, Part A, 2011, 155, 2609-2616.	1.2	47
46	Severe cleidocranial dysplasia can mimic hypophosphatasia. European Journal of Pediatrics, 2002, 161, 623-626.	2.7	46
47	Mutations in the cyclin family member FAM58A cause an X-linked dominant disorder characterized by syndactyly, telecanthus and anogenital and renal malformations. Nature Genetics, 2008, 40, 287-289.	21.4	45
48	TRPV4-pathy, a novel channelopathy affecting diverse systems. Journal of Human Genetics, 2010, 55, 400-402.	2.3	45
49	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	3.2	45
50	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. American Journal of Medical Genetics Part A, 1999, 85, 38-47.	2.4	44
51	Filamin A mutation is one cause of FG syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1876-1879.	1.2	44
52	Double heterozygosity for pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. American Journal of Medical Genetics Part A, 2001, 104, 140-146.	2.4	42
53	Identification of loss-of-function mutations of SLC35D1 in patients with Schneckenbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases. Journal of Medical Genetics, 2009, 46, 562-568.	3.2	41
54	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	3.2	39

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55	Chondrodysplasia punctata associated with maternal autoimmune diseases: Expanding the spectrum from systemic lupus erythematosus (SLE) to mixed connective tissue disease (MCTD) and scleroderma report of eight cases. American Journal of Medical Genetics, Part A, 2008, 146A, 3038-3053.	1.2	38
56	Mucolipidosis II presenting as severe neonatal hyperparathyroidism. European Journal of Pediatrics, 2005, 164, 236-243.	2.7	37
57	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€. American Journal of Human Genetics, 2017, 101, 815-823.	6.2	37
58	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. European Journal of Human Genetics, 2009, 17, 1141-1147.	2.8	33
59	StÃ¼veâ€“Wiedemann syndrome: longâ€term followâ€up and genetic heterogeneity. Clinical Genetics, 2010, 77, 266-272.	2.0	33
60	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. Genetics in Medicine, 2019, 21, 2734-2743.	2.4	33
61	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. American Journal of Medical Genetics Part A, 1998, 79, 103-107.	2.4	32
62	COL2A1â€“related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167.	1.2	32
63	A variant of Desbuquois dysplasia characterized by advanced carpal bone age, short metacarpals, and elongated phalanges: Report of seven cases. American Journal of Medical Genetics, Part A, 2010, 152A, 875-885.	1.2	32
64	Extracellular matrix and platelet function in patients with musculocontractural Ehlersâ€“Danlos syndrome caused by mutations in the CHST14 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1344-1354.	1.2	32
65	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2011, 89, 767-772.	6.2	31
66	A Genetic Approach to the Diagnosis of Skeletal Dysplasia. Clinical Orthopaedics and Related Research, 2002, 401, 32-38.	1.5	30
67	Fetal akinesia in metatropic dysplasia: The combined phenotype of chondrodysplasia and neuropathy?. American Journal of Medical Genetics, Part A, 2011, 155, 2860-2864.	1.2	30
68	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. JCI Insight, 2019, 4, .	5.0	30
69	Contiguous hemizygous deletion ofTBX5,TBX3, andRBM19 resulting in a combined phenotype of Holt-Oram and ulnar-mammary syndromes. American Journal of Medical Genetics, Part A, 2006, 140A, 1880-1886.	1.2	29
70	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. American Journal of Human Genetics, 2022, 109, 457-470.	6.2	29
71	Preselection of cases through expert clinical and radiological review significantly increases mutation detection rate in multiple epiphyseal dysplasia. European Journal of Human Genetics, 2007, 15, 150-154.	2.8	28
72	Novel de novo mutations in ZBTB20 in Primrose syndrome with congenital hypothyroidism. American Journal of Medical Genetics, Part A, 2016, 170, 1626-1629.	1.2	27

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73	Raine syndrome: A rare lethal osteosclerotic bone dysplasia. Prenatal diagnosis, autopsy, and neuropathological findings. American Journal of Medical Genetics, Part A, 2007, 143A, 3280-3285.	1.2	26
74	Evidence That Smith-McCort Dysplasia and Dyggve-Melchior-Clausen Dysplasia Are Allelic Disorders That Result from Mutations in a Gene on Chromosome 18q12. American Journal of Human Genetics, 2002, 71, 947-951.	6.2	23
75	Clinical and radiographic delineation of odontochondrodysplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 770-778.	1.2	23
76	Constitutional mismatch repair deficiencyâ€“associated brain tumors: report from the European C4CMMRD consortium. Neuro-Oncology Advances, 2019, 1, vdz033.	0.7	23
77	Leriâ€™s pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassingGDF6andSDC2and provides insight into systemic sclerosis pathogenesis. Annals of the Rheumatic Diseases, 2015, 74, 1249-1256.	0.9	22
78	Chondroitin SulfateN-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. Human Mutation, 2017, 38, 34-38.	2.5	22
79	Positive effects of an angiotensin II type 1 receptor antagonist in Camuratiâ€“Engelmann disease: A single case observation. American Journal of Medical Genetics, Part A, 2014, 164, 2667-2671.	1.2	21
80	Revisit of multiple epiphyseal dysplasia: Ethnic difference in genotypes and comparison of radiographic features linked to the COMP and MATN3 genes. American Journal of Medical Genetics, Part A, 2011, 155, 2669-2680.	1.2	20
81	Alâ€Awadiâ€“Raasâ€Rothschild (limb/pelvis/uterusâ€“hypoplasia/aplasia) syndrome and WNT7A mutations: Genetic homogeneity and nosological delineation. American Journal of Medical Genetics, Part A, 2011, 155, 332-336.	1.2	19
82	A family with a newelastingene mutation: broad clinical spectrum, including sudden cardiac death. Cardiology in the Young, 2011, 21, 62-65.	0.8	18
83	Clinical and radiological findings in Pallisterâ€“Killian syndrome. European Journal of Medical Genetics, 2012, 55, 167-172.	1.3	18
84	Eight years experience from a skeletal dysplasia referral center in a tertiary hospital in Southern India: A model for the diagnosis and treatment of rare diseases in a developing country. American Journal of Medical Genetics, Part A, 2014, 164, 2317-2323.	1.2	18
85	"Duplicate calcaneus": a rare developmental defect observed in several skeletal dysplasias. Pediatric Radiology, 2001, 31, 38-42.	2.0	17
86	Immunohistochemical Evaluation of Conjunctival Fibrillin-1 in Marfan Syndrome. JAMA Ophthalmology, 2006, 124, 205.	2.4	17
87	Challenges in managing genetic cancer risk: a long-term qualitative study of unaffected women carrying BRCA1/BRCA2 mutations. Genetics in Medicine, 2015, 17, 726-732.	2.4	17
88	Brief Report: Peripheral Osteolysis in Adults Linked to ASAH1 (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. Arthritis and Rheumatology, 2016, 68, 2323-2327.	5.6	17
89	Oto-palato-digital syndrome, type II: Report of three cases with further delineation of the chondro-osseous morphology. American Journal of Medical Genetics Part A, 2000, 95, 193-200.	2.4	16
90	Natural history and life-threatening complications in Myhre syndrome and review of the literature. European Journal of Pediatrics, 2016, 175, 1307-1315.	2.7	15

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91	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	2.5	15
92	The Shwachman-Bodian-Diamond syndrome gene mutations cause a neonatal form of spondylometaphysial dysplasia (SMD) resembling SMD Sedaghatian type. Journal of Medical Genetics, 2007, 44, e73-e73.	3.2	14
93	Fetal MR imaging of atelosteogenesis type II (AO-II). Pediatric Radiology, 2008, 38, 1345-1349.	2.0	14
94	Homozygosity for a novel truncating mutation confirms TBX15 deficiency as the cause of Cousin syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 3161-3165.	1.2	14
95	MMP13 mutations are the cause of recessive metaphyseal dysplasia, Spahr type. American Journal of Medical Genetics, Part A, 2014, 164, 1175-1179.	1.2	14
96	Petrified ears in a patient with Keutel syndrome: temporal bone CT findings. Pediatric Radiology, 2006, 36, 241-243.	2.0	13
97	Severe Peripheral Joint Laxity is a Distinctive Clinical Feature of Spondylodysplastic-Ehlers-Danlos Syndrome (EDS)-B4GALT7 and Spondylodysplastic-EDS-B3GALT6. Genes, 2019, 10, 799.	2.4	13
98	Fetus with two identical reciprocal translocations: Description of a rare complication of consanguinity. American Journal of Medical Genetics, Part A, 2006, 140A, 769-774.	1.2	12
99	Ligand Binding to the Collagen VI Receptor Triggers a Talin-to-RhoA Switch that Regulates Receptor Endocytosis. Developmental Cell, 2020, 53, 418-430.e4.	7.0	12
100	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	2.9	11
101	Is height important for quality of life in children with skeletal dysplasias?. European Journal of Medical Genetics, 2020, 63, 103816.	1.3	11
102	X-Linked dominant chondrodysplasia punctata: prenatal diagnosis and autopsy findings. Prenatal Diagnosis, 2006, 26, 1235-1240.	2.3	10
103	â€œIs NF1 a genetic skeletal disorder?â€â€”A response. American Journal of Medical Genetics, Part A, 2007, 143A, 2084-2084.	1.2	10
104	Phenotypic and molecular characterization of a novel case of dyssegmental dysplasia, Silverman-Handmaker type. European Journal of Medical Genetics, 2010, 53, 294-298.	1.3	10
105	Small patella syndrome: New clinical and molecular insights into a consistent phenotype. Clinical Genetics, 2017, 92, 676-678.	2.0	10
106	Does the clinical phenotype of mucolipidosis-IIIÎ³ differ from its Î±Î² counterpart?: supporting facts in a cohort of 18 patients. Clinical Dysmorphology, 2019, 28, 7-16.	0.3	10
107	Combined Lung and Liver Transplantation for Short Telomere Syndrome. Liver Transplantation, 2020, 26, 840-844.	2.4	10
108	The mildest form of campomelic dysplasia. American Journal of Medical Genetics, Part A, 2005, 132A, 113-113.	1.2	9

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109	A distinct form of spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)-leptodactylic type: radiological characteristics in seven new patients. Skeletal Radiology, 2009, 38, 803-811.	2.0	9
110	The Connective Tissue Disorder Associated with Recessive Variants in the SLC39A13 Zinc Transporter Gene (Spondylo-Dysplastic Ehlersâ€“Danlos Syndrome Type 3): Insights from Four Novel Patients and Follow-Up on Two Original Cases. Genes, 2020, 11, 420.	2.4	9
111	Stuve-Wiedemann syndrome with a novel mutation. BMJ Case Reports, 2015, 2015, bcr2015212032.	0.5	9
112	Axial spondylometaphyseal dysplasia: Additional reports. American Journal of Medical Genetics, Part A, 2011, 155, 2521-2528.	1.2	8
113	Acampomelic Form of Campomelic Dysplasia with SOX9 Missense Mutation. Indian Journal of Pediatrics, 2014, 81, 98-100.	0.8	8
114	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
115	New dysplasia or achondrogenesis type 1B? The importance of histology and molecular biology in delineating skeletal dysplasias. Pediatric Radiology, 2001, 31, 893-894.	2.0	7
116	A Diagnostic Approach to Skeletal Dysplasias. , 2003, , 375-IX.		7
117	Dyssegmental dysplasia, Silvermanâ€Handmaker type: prenatal ultrasound findings and molecular analysis. Prenatal Diagnosis, 2013, 33, 1039-1043.	2.3	7
118	A Diagnostic Approach to Skeletal Dysplasias. , 2012, , 403-437.		6
119	Physicians communicating with women at genetic risk of breast and ovarian cancer: Are we in the middle of the ford between contradictory messages and unshared decision making?. PLoS ONE, 2020, 15, e0240054.	2.5	6
120	Focal dermal hypoplasia (goltzâ€“gorlin syndrome): A new case with a novel variant in the PORCN gene (c.1250T>C:p.F417S) and unusual spinal anomaly. American Journal of Medical Genetics, Part A, 2013, 161, 1750-1754.	1.2	5
121	Genetic disorders of bone â€“ An historical perspective. Bone, 2017, 102, 1-4.	2.9	5
122	Confirmation of spondyloâ€epiâ€metaphyseal dysplasia with joint laxity, EXOC6B type. American Journal of Medical Genetics, Part A, 2018, 176, 2934-2935.	1.2	5
123	Clinical and Molecular Diagnosis of Osteocraniostenosis in Fetuses and Newborns: Prenatal Ultrasound, Clinical, Radiological and Pathological Features. Genes, 2022, 13, 261.	2.4	5
124	Non-invasive prenatal testing leading to a maternal diagnosis of Charcotâ€“Marieâ€“Tooth neuropathy. Journal of Human Genetics, 2020, 65, 1035-1038.	2.3	4
125	Immune deficiency, autoimmune disease and intellectual disability: A pleiotropic disorder caused by biallelic variants in the <scp>TPP2</scp> gene. Clinical Genetics, 2021, 99, 780-788.	2.0	4
126	Update Swiss guideline for counselling and testing for predisposition to breast, ovarian, pancreatic and prostate cancer. Swiss Medical Weekly, 2021, 151, w30038.	1.6	4

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127	Osteogenesis imperfecta: towards an individualised interdisciplinary care strategy to improve physical activity and quality of life. Swiss Medical Weekly, 2020, 150, w20285.	1.6	4
128	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	1.2	3
129	A familial case of trichoâ€rhinoâ€phalangeal syndrome type III with a novel missense mutation in exon 6 of the TRPS1 gene. Journal of the European Academy of Dermatology and Venereology, 2010, 24, 612-614.	2.4	2
130	Chondrodysplasias. , 2013, , 1-45.		2
131	Cancer surveillance in children with Ollier Disease and Maffucci Syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1338-1340.	1.2	2
132	Partial tetrasomy with triplication of chromosome (5) (p14â€p15.33) in a patient with severe multiple congenital anomalies. American Journal of Medical Genetics Part A, 1998, 79, 103-107.	2.4	2
133	IGF-I Receptor Mutations and Intrauterine and Postnatal Growth Retardation. New England Journal of Medicine, 2004, 350, 1362-1363.	27.0	1
134	New topics in the skeletal dysplasias. , 2012, 160C, 143-144.		1
135	A Novel Talin-to-RhoA Switch Mechanism Upon Ligand Binding of the Collagen VI Receptor CMG2. SSRN Electronic Journal, 0, , .	0.4	1
136	Congenital Joint Dislocations Caused by Carbohydrate Sulfotransferase 3 Deficiency in Recessive Larsen Syndrome and Humero-Spinal Dysostosis. American Journal of Human Genetics, 2008, 83, 293.	6.2	0
137	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. American Journal of Human Genetics, 2009, 85, 420.	6.2	0
138	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2012, 90, 170.	6.2	0
139	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0
140	Sojourner in a foreign land. American Journal of Medical Genetics, Part A, 2016, 170, 2594-2595.	1.2	0
141	The Bone in Genetic and Metabolic Diseases: A Practical Approach. , 2017, , 371-380.		0
142	Complex cranio-vertebral malformation: disruption sequence or iniencephaly?. Clinical Dysmorphology, 2018, 27, 105-108.	0.3	0