

Sheila L Unger

List of Publications by Year in descending order

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142
papers

6,904
citations

57631

44
h-index

69108

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.	0.7	573
2	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	0.7	453
3	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	0.7	431
4	Nosology and classification of genetic skeletal disorders: 2006 revision. American Journal of Medical Genetics, Part A, 2007, 143A, 1-18.	0.7	301
5	The Zinc Transporter SLC39A13/ZIP13 Is Required for Connective Tissue Development; Its Involvement in BMP/TGF- β Signaling Pathways. PLoS ONE, 2008, 3, e3642.	1.1	240
6	Mutations in the TGF β 2 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
7	Null Leukemia Inhibitory Factor Receptor (LIFR) Mutations in StÅ¼ave-Wiedemann/Schwartz-Jampel Type 2 Syndrome. American Journal of Human Genetics, 2004, 74, 298-305.	2.6	162
8	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	9.4	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.	9.4	125
11	Cortical-Bone Fragility â€” Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	13.9	119
12	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	2.6	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.	2.6	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.	2.6	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.	1.1	104
16	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	1.5	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.	1.1	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.	2.6	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.	2.6	88
20	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and <i>WISP3</i> mutations in 63 affected individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 217-229.	0.7	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.	3.3	73
22	TRPV4-associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	0.7	71
23	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. Human Genetics, 2011, 129, 497-502.	1.8	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.	0.7	67
25	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	1.1	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.	0.7	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.	1.6	65
28	Multiple epiphyseal dysplasia: radiographic abnormalities correlated with genotype. Pediatric Radiology, 2001, 31, 10-18.	1.1	61
29	Mutations in <i>LONP1</i> , a mitochondrial matrix protease, cause CODAS syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1501-1509.	0.7	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.	2.6	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.	0.7	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.	1.5	58
33	Evolutionary Comparison Provides Evidence for Pathogenicity of RMRP Mutations. PLoS Genetics, 2005, 1, e47.	1.5	57
34	Defective C-propeptides of the Pro α 2(I) Chain of Type I Procollagen Impede Molecular Assembly and Result in Osteogenesis Imperfecta. Journal of Biological Chemistry, 2008, 283, 16061-16067.	1.6	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.	1.4	56
36	Spondyloepiphyseal dysplasia, Maroteaux type (pseudo-Morquio syndrome type 2), and parastremmatic dysplasia are caused by <i>TRPV4</i> mutations. American Journal of Medical Genetics, Part A, 2010, 152A, 1443-1449.	0.7	56

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37	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	1.4	56
38	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. <i>Nature Communications</i> , 2017, 8, 15861.	5.8	56
39	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 168-178.	2.6	54
40	Mental Retardation and Abnormal Skeletal Development (Dyggve-Melchior-Clausen Dysplasia) Due to Mutations in a Novel, Evolutionarily Conserved Gene. <i>American Journal of Human Genetics</i> , 2003, 72, 419-428.	2.6	53
41	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. <i>American Journal of Human Genetics</i> , 2013, 92, 137-143.	2.6	53
42	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. <i>Nature</i> , 2021, 592, 93-98.	13.7	53
43	Current Care and Investigational Therapies in Achondroplasia. <i>Current Osteoporosis Reports</i> , 2017, 15, 53-60.	1.5	50
44	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 395-400.	1.4	48
45	Whole-exome sequencing detects somatic mutations of <i>IDH1</i> in metaphyseal chondromatosis with <i>D</i> -2-hydroxyglutaric aciduria (MC-HGA). <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2609-2616.	0.7	47
46	Severe cleidocranial dysplasia can mimic hypophosphatasia. <i>European Journal of Pediatrics</i> , 2002, 161, 623-626.	1.3	46
47	Mutations in the cyclin family member FAM58A cause an X-linked dominant disorder characterized by syndactyly, telecanthus and anogenital and renal malformations. <i>Nature Genetics</i> , 2008, 40, 287-289.	9.4	45
48	TRPV4-pathy, a novel channelopathy affecting diverse systems. <i>Journal of Human Genetics</i> , 2010, 55, 400-402.	1.1	45
49	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. <i>Journal of Medical Genetics</i> , 2011, 48, 417-421.	1.5	45
50	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. <i>American Journal of Medical Genetics Part A</i> , 1999, 85, 38-47.	2.4	44
51	<i>Filamin A</i> mutation is one cause of FG syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1876-1879.	0.7	44
52	Double heterozygosity for pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. <i>American Journal of Medical Genetics Part A</i> , 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. <i>Journal of Medical Genetics</i> , 2009, 46, 562-568.	1.5	41
54	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. <i>Journal of Medical Genetics</i> , 2011, 48, 32-37.	1.5	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. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3038-3053.	0.7	38
56	Mucopolipidosis II presenting as severe neonatal hyperparathyroidism. <i>European Journal of Pediatrics</i> , 2005, 164, 236-243.	1.3	37
57	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". <i>American Journal of Human Genetics</i> , 2017, 101, 815-823.	2.6	37
58	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. <i>European Journal of Human Genetics</i> , 2009, 17, 1141-1147.	1.4	33
59	StÃ¼ve-Wiedemann syndrome: long-term follow-up and genetic heterogeneity. <i>Clinical Genetics</i> , 2010, 77, 266-272.	1.0	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. <i>Genetics in Medicine</i> , 2019, 21, 2734-2743.	1.1	33
61	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. <i>American Journal of Medical Genetics Part A</i> , 1998, 79, 103-107.	2.4	32
62	COL2A1-related skeletal dysplasias with predominant metaphyseal involvement. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 161-167.	0.7	32
63	A variant of Desbuquois dysplasia characterized by advanced carpal bone age, short metacarpals, and elongated phalanges: Report of seven cases. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 875-885.	0.7	32
64	Extracellular matrix and platelet function in patients with musculocontractural Ehlers-Danlos syndrome caused by mutations in the <i>CHST14</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1344-1354.	0.7	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. <i>American Journal of Human Genetics</i> , 2011, 89, 767-772.	2.6	31
66	A Genetic Approach to the Diagnosis of Skeletal Dysplasia. <i>Clinical Orthopaedics and Related Research</i> , 2002, 401, 32-38.	0.7	30
67	Fetal akinesia in metatropic dysplasia: The combined phenotype of chondrodysplasia and neuropathy?. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2860-2864.	0.7	30
68	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. <i>JCI Insight</i> , 2019, 4, .	2.3	30
69	Contiguous hemizygous deletion of TBX5, TBX3, and RBM19 resulting in a combined phenotype of Holt-Oram and ulnar-mammary syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1880-1886.	0.7	29
70	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. <i>American Journal of Human Genetics</i> , 2022, 109, 457-470.	2.6	29
71	Preselection of cases through expert clinical and radiological review significantly increases mutation detection rate in multiple epiphyseal dysplasia. <i>European Journal of Human Genetics</i> , 2007, 15, 150-154.	1.4	28
72	Novel de novo mutations in <i>ZBTB20</i> in Primrose syndrome with congenital hypothyroidism. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1626-1629.	0.7	27

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73	Raine syndrome: A rare lethal osteosclerotic bone dysplasia. Prenatal diagnosis, autopsy, and neuropathological findings. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3280-3285.	0.7	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. <i>American Journal of Human Genetics</i> , 2002, 71, 947-951.	2.6	23
75	Clinical and radiographic delineation of odontochondrodysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 770-778.	0.7	23
76	Constitutional mismatch repair deficiency-associated brain tumors: report from the European C4MMRD consortium. <i>Neuro-Oncology Advances</i> , 2019, 1, vdz033.	0.4	23
77	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing <i>GDF6</i> and <i>SDC2</i> and provides insight into systemic sclerosis pathogenesis. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1249-1256.	0.5	22
78	Chondroitin Sulfate <i>N</i> -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
79	Positive effects of an angiotensin II type 1 receptor antagonist in Camurati-Engelmann disease: A single case observation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2667-2671.	0.7	21
80	Revisit of multiple epiphyseal dysplasia: Ethnic difference in genotypes and comparison of radiographic features linked to the COMP and MATN3 genes. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2669-2680.	0.7	20
81	Al-Awadi-Raas-Rothschild (limb/pelvis/uterus hypoplasia/aplasia) syndrome and <i>WNT7A</i> mutations: Genetic homogeneity and nosological delineation. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 332-336.	0.7	19
82	A family with a new <i>elastin</i> gene mutation: broad clinical spectrum, including sudden cardiac death. <i>Cardiology in the Young</i> , 2011, 21, 62-65.	0.4	18
83	Clinical and radiological findings in Pallister-Killian syndrome. <i>European Journal of Medical Genetics</i> , 2012, 55, 167-172.	0.7	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. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2317-2323.	0.7	18
85	"Duplicate calcaneus": a rare developmental defect observed in several skeletal dysplasias. <i>Pediatric Radiology</i> , 2001, 31, 38-42.	1.1	17
86	Immunohistochemical Evaluation of Conjunctival Fibrillin-1 in Marfan Syndrome. <i>JAMA Ophthalmology</i> , 2006, 124, 205.	2.6	17
87	Challenges in managing genetic cancer risk: a long-term qualitative study of unaffected women carrying BRCA1/BRCA2 mutations. <i>Genetics in Medicine</i> , 2015, 17, 726-732.	1.1	17
88	Brief Report: Peripheral Osteolysis in Adults Linked to <i>ASAHI</i> (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. <i>Arthritis and Rheumatology</i> , 2016, 68, 2323-2327.	2.9	17
89	Oto-palato-digital syndrome, type II: Report of three cases with further delineation of the chondro-osseous morphology. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 193-200.	2.4	16
90	Natural history and life-threatening complications in Myhre syndrome and review of the literature. <i>European Journal of Pediatrics</i> , 2016, 175, 1307-1315.	1.3	15

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91	CSGALNACT1 congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. <i>Human Mutation</i> , 2020, 41, 655-667.	1.1	15
92	The Shwachman-Bodian-Diamond syndrome gene mutations cause a neonatal form of spondylometaphyseal dysplasia (SMD) resembling SMD Sedaghatian type. <i>Journal of Medical Genetics</i> , 2007, 44, e73-e73.	1.5	14
93	Fetal MR imaging of atelosteogenesis type II (AO-II). <i>Pediatric Radiology</i> , 2008, 38, 1345-1349.	1.1	14
94	Homozygosity for a novel truncating mutation confirms <i>TBX15</i> deficiency as the cause of Cousin syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 3161-3165.	0.7	14
95	<i>MMP13</i> mutations are the cause of recessive metaphyseal dysplasia, Spahr type. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1175-1179.	0.7	14
96	Petrified ears in a patient with Keutel syndrome: temporal bone CT findings. <i>Pediatric Radiology</i> , 2006, 36, 241-243.	1.1	13
97	Severe Peripheral Joint Laxity is a Distinctive Clinical Feature of Spondylodysplastic-Ehlers-Danlos Syndrome (EDS)-B4GALT7 and Spondylodysplastic-EDS-B3GALT6. <i>Genes</i> , 2019, 10, 799.	1.0	13
98	Fetus with two identical reciprocal translocations: Description of a rare complication of consanguinity. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 769-774.	0.7	12
99	Ligand Binding to the Collagen VI Receptor Triggers a Talin-to-RhoA Switch that Regulates Receptor Endocytosis. <i>Developmental Cell</i> , 2020, 53, 418-430.e4.	3.1	12
100	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. <i>Bone</i> , 2019, 120, 354-363.	1.4	11
101	Is height important for quality of life in children with skeletal dysplasias?. <i>European Journal of Medical Genetics</i> , 2020, 63, 103816.	0.7	11
102	X-Linked dominant chondrodysplasia punctata: prenatal diagnosis and autopsy findings. <i>Prenatal Diagnosis</i> , 2006, 26, 1235-1240.	1.1	10
103	Does NF1 a genetic skeletal disorder? A response. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2084-2084.	0.7	10
104	Phenotypic and molecular characterization of a novel case of dyssegmental dysplasia, Silverman-Handmaker type. <i>European Journal of Medical Genetics</i> , 2010, 53, 294-298.	0.7	10
105	Small patella syndrome: New clinical and molecular insights into a consistent phenotype. <i>Clinical Genetics</i> , 2017, 92, 676-678.	1.0	10
106	Does the clinical phenotype of mucopolidosis-III differ from its counterpart?: supporting facts in a cohort of 18 patients. <i>Clinical Dysmorphology</i> , 2019, 28, 7-16.	0.1	10
107	Combined Lung and Liver Transplantation for Short Telomere Syndrome. <i>Liver Transplantation</i> , 2020, 26, 840-844.	1.3	10
108	The mildest form of campomelic dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 113-113.	0.7	9

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109	A distinct form of spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)-leptodactylic type: radiological characteristics in seven new patients. <i>Skeletal Radiology</i> , 2009, 38, 803-811.	1.2	9
110	The Connective Tissue Disorder Associated with Recessive Variants in the SLC39A13 Zinc Transporter Gene (Spondylo-Dysplastic Ehlers-Danlos Syndrome Type 3): Insights from Four Novel Patients and Follow-Up on Two Original Cases. <i>Genes</i> , 2020, 11, 420.	1.0	9
111	Stuve-Wiedemann syndrome with a novel mutation. <i>BMJ Case Reports</i> , 2015, 2015, bcr2015212032.	0.2	9
112	Axial spondylometaphyseal dysplasia: Additional reports. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2521-2528.	0.7	8
113	Acampomelic Form of Campomelic Dysplasia with SOX9 Missense Mutation. <i>Indian Journal of Pediatrics</i> , 2014, 81, 98-100.	0.3	8
114	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
115	New dysplasia or achondrogenesis type 1B? The importance of histology and molecular biology in delineating skeletal dysplasias. <i>Pediatric Radiology</i> , 2001, 31, 893-894.	1.1	7
116	A Diagnostic Approach to Skeletal Dysplasias. , 2003, , 375-IX.		7
117	Dyssegmental dysplasia, Silverman-Handmaker type: prenatal ultrasound findings and molecular analysis. <i>Prenatal Diagnosis</i> , 2013, 33, 1039-1043.	1.1	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?. <i>PLoS ONE</i> , 2020, 15, e0240054.	1.1	6
120	Focal dermal hypoplasia (goltz-gorlin syndrome): A new case with a novel variant in the <i>PORCN</i> gene (c.1250T>C;p.F417S) and unusual spinal anomaly. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1750-1754.	0.7	5
121	Genetic disorders of bone – An historical perspective. <i>Bone</i> , 2017, 102, 1-4.	1.4	5
122	Confirmation of spondyloepimetaphyseal dysplasia with joint laxity, <i>EXOC6B</i> type. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2934-2935.	0.7	5
123	Clinical and Molecular Diagnosis of Osteocraniostenosis in Fetuses and Newborns: Prenatal Ultrasound, Clinical, Radiological and Pathological Features. <i>Genes</i> , 2022, 13, 261.	1.0	5
124	Non-invasive prenatal testing leading to a maternal diagnosis of Charcot-Marie-Tooth neuropathy. <i>Journal of Human Genetics</i> , 2020, 65, 1035-1038.	1.1	4
125	Immune deficiency, autoimmune disease and intellectual disability: A pleiotropic disorder caused by biallelic variants in the <i>TPP2</i> gene. <i>Clinical Genetics</i> , 2021, 99, 780-788.	1.0	4
126	Update Swiss guideline for counselling and testing for predisposition to breast, ovarian, pancreatic and prostate cancer. <i>Swiss Medical Weekly</i> , 2021, 151, w30038.	0.8	4

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127	Osteogenesis imperfecta: towards an individualised interdisciplinary care strategy to improve physical activity and quality of life. <i>Swiss Medical Weekly</i> , 2020, 150, w20285.	0.8	4
128	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 517-527.	0.7	3
129	A familial case of tricho-rhino-phalangeal syndrome type III with a novel missense mutation in exon 6 of the <i>TRPS1</i> gene. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2010, 24, 612-614.	1.3	2
130	Chondrodysplasias. , 2013, , 1-45.		2
131	Cancer surveillance in children with Ollier Disease and Maffucci Syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1338-1340.	0.7	2
132	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. , 1998, 79, 103.		2
133	IGF-I Receptor Mutations and Intrauterine and Postnatal Growth Retardation. <i>New England Journal of Medicine</i> , 2004, 350, 1362-1363.	13.9	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. <i>SSRN Electronic Journal</i> , 0, , .	0.4	1
136	Congenital Joint Dislocations Caused by Carbohydrate Sulfotransferase 3 Deficiency in Recessive Larsen Syndrome and Humero-Spinal Dysostosis. <i>American Journal of Human Genetics</i> , 2008, 83, 293.	2.6	0
137	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 420.	2.6	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. <i>American Journal of Human Genetics</i> , 2012, 90, 170.	2.6	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. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2594-2595.	0.7	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?. <i>Clinical Dysmorphology</i> , 2018, 27, 105-108.	0.1	0