Andrea Superti-Furga

List of Publications by Year
in descending order

334 papers	20,530 citations	13099 68 h-index	14208 128 g-index
351 all docs	351 docs citations	351 times ranked	19762 citing authors

#	Article	IF	Citations
1	A monoallelic<scp>SEC23A</scp>variant<scp>E599K</scp>associated with<scp>cranioâ€lenticuloâ€sutural</scp>dysplasia. American Journal of Medical Genetics, Part A, 2022, 188, 319-325.	1.2	3
2	Clinical and Molecular Diagnosis of Osteocraniostenosis in Fetuses and Newborns: Prenatal Ultrasound, Clinical, Radiological and Pathological Features. Genes, 2022, 13, 261.	2.4	5
3	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
4	SCN5A Overlap Syndromes: an open-minded approach. Heart Rhythm, 2022, , .	0.7	2
5	Clinical and Genetic Findings in a Series of Eight Families with Arthrogryposis. Genes, 2022, 13, 29.	2.4	6
6	Identification of Disease Gene for Camurati-Engelmann Disease, Type II. Bone Reports, 2022, 16, 101561.	0.4	0
7	Biallelic variants in ZNF526 cause a severe neurodevelopmental disorder with microcephaly, bilateral cataract, epilepsy and simplified gyration. Journal of Medical Genetics, 2021, , jmedgenet-2020-107430.	3.2	5
8	Cancer surveillance in children with Ollier Disease and Maffucci Syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1338-1340.	1.2	2
9	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
10	Syndromic disorders caused by gain-of-function variants in KCNH1, KCNK4, and KCNN3â€”a subgroup of K+ channelopathies. European Journal of Human Genetics, 2021, 29, 1384-1395.	2.8	21
11	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
12	Spinal cerebrotendinous xanthomatosis: A case report and literature review. Molecular Genetics and Metabolism Reports, 2021, 26, 100719.	1.1	8
13	Improvement of the skeletal phenotype in a mouse model of diastrophic dysplasia after postnatal treatment with N-acetylcysteine. Biochemical Pharmacology, 2021, 185, 114452.	4.4	10
14	<scp>CNOT2</scp> haploinsufficiency in a 40â€yearâ€old man with intellectual disability, autism, and seizures. American Journal of Medical Genetics, Part A, 2021, 185, 2602-2606.	1.2	3
15	NGS-Based Diagnosis of Treatable Neurogenetic Disorders in Adults: Opportunities and Challenges. Genes, 2021, 12, 695.	2.4	5
16	Case Report: A Rare Truncating Variant of the CFHR5 Gene in IgA Nephropathy. Frontiers in Genetics, 2021, 12, 529236.	2.3	3
17	Classical homocystinuria, is it safe to exercise?. Molecular Genetics and Metabolism Reports, 2021, 27, 100746.	1.1	1
18	Biallelic deep intronic variant c.5457+81T>A in TRIP11 causes loss of function and results in achondrogenesis 1A. Human Mutation, 2021, 42, 1005-1014.	2.5	3

#	Article	IF	Citations
19	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
20	Oâ€™Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. Journal of Medical Genetics, 2021, , jmedgenet-2020-107470.	3.2	4
21	Homozygous GLI3 variants observed in three unrelated patients presenting with syndromic polydactyly. American Journal of Medical Genetics, Part A, 2021, 185, 3831-3837.	1.2	0
22	Elevated lactate in Mauriac syndrome: still a mystery. BMC Endocrine Disorders, 2021, 21, 172.	2.2	1
23	CNV Detection from Exome Sequencing Data in Routine Diagnostics of Rare Genetic Disorders: Opportunities and Limitations. Genes, 2021, 12, 1427.	2.4	21
24	Agenesis of the Corpus Callosum with Facial Dysmorphism and Intellectual Disability in Sibs Associated with Compound Heterozygous KDM5B Variants. Genes, 2021, 12, 1397.	2.4	1
25	Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. Scientific Reports, 2021, 11, 19332.	3.3	2
26	The fate of orally administered sialic acid: First insights from patients with N-acetylneuraminic acid synthase deficiency and control subjects. Molecular Genetics and Metabolism Reports, 2021, 28, 100777.	1.1	7
27	AutoMap is a high performance homozygosity mapping tool using next-generation sequencing data. Nature Communications, 2021, 12, 518.	12.8	68
28	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
29	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
30	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	2.5	15
31	Exploring the Genetic Landscape of Retinal Diseases in North-Western Pakistan Reveals a High Degree of Autozygosity and a Prevalent Founder Mutation in ABCA4. Genes, 2020, 11, 12.	2.4	13
32	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
33	Clouds over IMD? Perspectives for inherited metabolic diseases in adults from a retrospective cohort study in two Swiss adult metabolic clinics. Orphanet Journal of Rare Diseases, 2020, 15, 210.	2.7	14
34	An Alu-mediated duplication in NMNAT1, involved in NAD biosynthesis, causes a novel syndrome, SHILCA, affecting multiple tissues and organs. Human Molecular Genetics, 2020, 29, 2250-2260.	2.9	14
35	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
36	Clinical aspects of Hyaline Fibromatosis Syndrome and identification of a novel mutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1203.	1.2	13

#	Article	IF	Citations
37	Childhood neurodegeneration associated with a specific UBTF variant: a new case report and review of the literature. BMC Neurology, 2020, 20, 17.	1.8	15
38	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
39	Skeletal Dysplasias Caused by Sulfation Defects. International Journal of Molecular Sciences, 2020, 21, 2710.	4.1	18
40	Collagen Type 1 and Osteogenesis Imperfecta. , 2020, , 125-129.		0
41	Hepatosplenomegaly, pneumopathy, bone changes and fronto-temporal dementia: Niemannâ€“Pick type B and SQSTM1-associated Pagetâ€™s disease in the same individual. Journal of Bone and Mineral Metabolism, 2019, 37, 378-383.	2.7	1
42	Bone and connective tissue disorders caused by defects in glycosaminoglycan biosynthesis: a panoramic view. FEBS Journal, 2019, 286, 3008-3032.	4.7	37
43	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
44	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
45	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
46	Peripheral neuropathy and cognitive impairment associated with a novel monoallelic <scp>HARS</scp> variant. Annals of Clinical and Translational Neurology, 2019, 6, 1072-1080.	3.7	15
47	A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2019, 40, 177-181.	1.2	10
48	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
49	AB1035â€…MAFB-VARIANTS IN MULTICENTRIC CARPOTARSAL OSTEOLYSIS WITH NEPHROPATHY DO NOT SEEM TO AFFECT SERUM C1Q CONCENTRATION. , 2019, , .		0
50	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. American Journal of Human Genetics, 2019, 105, 1294-1301.	6.2	17
51	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	2.9	11
52	Progressive pseudorheumatoid dysplasia: a rare childhood disease. Rheumatology International, 2019, 39, 441-452.	3.0	22
53	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. JCI Insight, 2019, 4, .	5.0	30
54	Dysostosen. Springer Reference Medizin, 2019, , 1-12.	0.0	0

#	Article	IF	Citations
55	When Materials Are at Fault: The Skeletal Collagens, Osteogenesis Imperfecta and Chondrodysplasias. , 2019, , 255-266.		0
56	Complex cranio-vertebral malformation: disruption sequence or iniencephaly?. Clinical Dysmorphology, 2018, 27, 105-108.	0.3	0
57	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
58	A novel in-frame deletion in ZMPSTE24 is associated with autosomal recessive acrogeria (Gottron) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	0.3	4
59	Prominent and elongated coccyx, a new manifestation of KBG syndrome associated with novel mutation in ANKRD11. American Journal of Medical Genetics, Part A, 2018, 176, 1991-1995.	1.2	10
60	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
61	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	1.2	24
62	The multiple faces of artwork diagnoses. Lancet Neurology, The, 2017, 16, 417.	10.2	4
63	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. Nature Communications, 2017, 8, 15861.	12.8	56
64	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	6.2	90
65	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
66	Genetic disorders of bone â€“ An historical perspective. Bone, 2017, 102, 1-4.	2.9	5
67	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
68	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
69	The Bone in Genetic and Metabolic Diseases: A Practical Approach. , 2017, , 371-380.		0
70	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
71	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
72	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254

#	Article	IF	Citations
73	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
74	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
75	Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. American Journal of Human Genetics, 2016, 99, 392-406.	6.2	52
76	Bone Formation and the Wnt Signaling Pathway. New England Journal of Medicine, 2016, 375, 1902-1903.	27.0	19
77	Bisphosphonates in multicentric osteolysis, nodulosis and arthropathy (MONA) spectrum disorder â€“ an alternative therapeutic approach. Scientific Reports, 2016, 6, 34017.	3.3	20
78	BGN Mutations in X-Linked Spondyloepimetaphyseal Dysplasia. American Journal of Human Genetics, 2016, 98, 1243-1248.	6.2	29
79	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	27.0	119
80	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
81	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
82	Mutations in LONP1, a mitochondrial matrix protease, cause CODAS syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1501-1509.	1.2	61
83	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
84	Analysis of the genetic basis of periodic fever with aphthous stomatitis, pharyngitis and cervical adenitis (PFAPA) syndrome. Scientific Reports, 2015, 5, 10200.	3.3	70
85	Significant clinical benefits of molecular studies in the skeletal dysplasias. American Journal of Medical Genetics, Part A, 2015, 167, 476-477.	1.2	1
86	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
87	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	1.2	453
88	Six additional cases of SEDC due to the same and recurrent R989C mutation in the COL2A1 geneâ€"the clinical and radiological followâ€up. American Journal of Medical Genetics, Part A, 2015, 167, 894-901.	1.2	8
89	Buried in the Middle but Guilty: Intronic Mutations in the TCIRG1 Gene Cause Human Autosomal Recessive Osteopetrosis. Journal of Bone and Mineral Research, 2015, 30, 1814-1821.	2.8	39
90	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. Human Molecular Genetics, 2015, 24, 1410-1419.	2.9	70

#	Article	IF	Citations
91	N-acetylcysteine treatment ameliorates the skeletal phenotype of a mouse model of diastrophic dysplasia. Human Molecular Genetics, 2015, 24, 5570-5580.	2.9	22
92	Multiple sulfatase deficiency with neonatal manifestation. Italian Journal of Pediatrics, 2014, 40, 86.	2.6	13
93	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
94	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
95	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0
96	Molecular pathogenesis of Spondylocheirodysplastic Ehlersâ€Ðanlos syndrome caused by mutant ZIP13 proteins. EMBO Molecular Medicine, 2014, 6, 1028-1042.	6.9	56
97	Acampomelic Form of Campomelic Dysplasia with SOX9 Missense Mutation. Indian Journal of Pediatrics, 2014, 81, 98-100.	0.8	8
98	Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 2014, 59, 122-126.	2.9	26
99	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
100	Angeborene EntwicklungsstÃ¶rungen des Skeletts. , 2014, , 1877-1911.		0
101	Propionic acidemia: clinical course and outcome in 55 pediatric and adolescent patients. Orphanet Journal of Rare Diseases, 2013, 8, 6.	2.7	138
102	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
103	3-M syndrome associated with growth hormone deficiency: 18Âyear follow-up of a patient. Italian Journal of Pediatrics, 2013, 39, 21.	2.6	24
104	Multiple tumor types including leiomyoma and Wilms tumor in a patient with Gorlin syndrome due to 9q22.3 microdeletion encompassing the PTCH1 and FANCâ€C loci. American Journal of Medical Genetics, Part A, 2013, 161, 2894-2901.	1.2	17
105	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
106	In-Depth Analysis of Hyaline Fibromatosis Syndrome Frameshift Mutations at the Same Site Reveal the Necessity of Personalized Therapy. Human Mutation, 2013, 34, 1005-1017.	2.5	14
107	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
108	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	6.2	114

#	Article	IF	Citations
109	Exome Sequencing Identifies INPPL1 Mutations as a Cause of Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 144-149.	6.2	44
110	Prenatal presentation and postnatal evolution of a patient with Jansen metaphyseal dysplasia with a novel missense mutation in PTH1R. American Journal of Medical Genetics, Part A, 2013, 161, 2614-2619.	1.2	11
111	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	3.2	127
112	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
113	Longâ€term followâ€up of four patients with langerâ€“giedion syndrome: Clinical course and complications. American Journal of Medical Genetics, Part A, 2013, 161, 2216-2225.	1.2	17
114	The dark sides of capillary morphogenesis gene 2. EMBO Journal, 2012, 31, 3-13.	7.8	71
115	An additional family with association of hereditary thrombocytosis and transverse limb deficiency: Confirmation of a rare clinical spectrum. American Journal of Medical Genetics, Part A, 2012, 158A, 3211-3213.	1.2	2
116	Metaphyseal chondromatosis combined with D-2-hydroxyglutaric aciduria in four patients. Skeletal Radiology, 2012, 41, 1479-1487.	2.0	12
117	A Diagnostic Approach to Skeletal Dysplasias. , 2012, , 403-437.		6
118	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	2.5	74
119	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
120	Simpsonâ€“Golabiâ€“Behmel syndrome type 1 in a 27â€week macrosomic preterm newborn: The diagnostic value of rib malformations and index nail and finger hypoplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2245-2249.	1.2	12
121	Enchondromatosis revisited: New classification with molecular basis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 154-164.	1.6	31
122	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
123	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	1.6	71
124	New topics in the skeletal dysplasias. , 2012, 160C, 143-144.		1
125	Severe neurologic manifestations from cervical spine instability in spondylo-megaepiphyseal-metaphyseal dysplasia. , 2012, 160C, 230-237.		10
126	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192

#	Article	IF	Citations
127	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
128	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
129	Mutation analysis in 54 propionic acidemia patients. Journal of Inherited Metabolic Disease, 2012, 35, 51-63.	3.6	41
130	Propionic acidemia: neonatal versus selective metabolic screening. Journal of Inherited Metabolic Disease, 2012, 35, 41-49.	3.6	69
131	Bone Dysplasias. , 2012, , .		42
132	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
133	Deletion of human GP1BB and SEPT5 is associated with Bernard-Soulier syndrome, platelet secretion defect, polymicrogyria, and developmental delay. Thrombosis and Haemostasis, 2011, 106, 475-483.	3.4	37
134	Clinical and molecular characterization of Diastrophic Dysplasia in the Portuguese population. Clinical Genetics, 2011, 80, 550-557.	2.0	13
135	Circulating matrix Î³-carboxyglutamate protein (MGP) species are refractory to vitaminÂK treatment in a new case of Keutel syndrome. Journal of Thrombosis and Haemostasis, 2011, 9, 1225-1235.	3.8	29
136	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	21.4	151
137	Hyperpyrexia resulting in encephalopathy in a 14-month-old patient with cblC disease. Brain and Development, 2011, 33, 432-436.	1.1	123
138	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
139	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
140	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
141	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. Human Genetics, 2011, 129, 497-502.	3.8	68
142	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
143	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	1.2	573
144	Axial spondylometaphyseal dysplasia: Additional reports. American Journal of Medical Genetics, Part A, 2011, 155, 2521-2528.	1.2	8

#	Article	IF	Citations
145	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
146	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
147	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
148	Hyaline Fibromatosis Syndrome inducing mutations in the ectodomain of anthrax toxin receptor 2 can be rescued by proteasome inhibitors. EMBO Molecular Medicine, 2011, 3, 208-221.	6.9	45
149	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
150	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	3.2	39
151	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	3.5	104
152	Mutations in the Gene Encoding the RER Protein FKBP65 Cause Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 86, 551-559.	6.2	278
153	Recessive multiple epiphyseal dysplasia (rMED) with homozygosity for C653S mutation in the DTDST gene - Phenotype, molecular diagnosis and surgical treatment of habitual dislocation of multilayered patella: Case report. BMC Musculoskeletal Disorders, 2010, 11, 110.	1.9	19
154	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
155	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
156	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
157	Current themes in molecular pediatrics: molecular medicine and its applications. Italian Journal of Pediatrics, 2010, 36, 20.	2.6	1
158	StÃ¼veâ€“Wiedemann syndrome: longâ€term followâ€up and genetic heterogeneity. Clinical Genetics, 2010, 77, 266-272.	2.0	33
159	A dominant mesomelic dysplasia associated with a 1.0-Mb microduplication of HOXD gene cluster at 2q31.1. Journal of Medical Genetics, 2010, 47, 638-639.	3.2	20
160	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	27.0	122
161	Novel Homozygous Mutation (c.175delG) in Platelet GlycoproteinITGA2BGene as Cause of Glanzmann's Thrombasthenia Type I. Klinische Padiatrie, 2010, 222, 150-153.	0.6	6
162	TRPV4-pathy, a novel channelopathy affecting diverse systems. Journal of Human Genetics, 2010, 55, 400-402.	2.3	45

#	Article	IF	Citations
163	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
164	Fatal adult-onset antibody deficiency syndrome in a patient with cartilage hair hypoplasia. Human Immunology, 2010, 71, 916-919.	2.4	8
165	Defective proteoglycan sulfation of the growth plate zones causes reduced chondrocyte proliferation via an altered Indian hedgehog signalling. Matrix Biology, 2010, 29, 453-460.	3.6	44
166	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
167	Difficulties in Diagnosis and Treatment of 5Î±-Reductase Type 2 Deficiency in a Newborn with 46,XY DSD. Hormone Research in Paediatrics, 2010, 74, 67-71.	1.8	53
168	Lepirudin Treatment in a Girl with Iliac Vein Thrombosis, Severe Pulmonary Embolism and Suspected Heparin-induced Thrombocytopenia (HIT) II. Klinische Padiatrie, 2009, 221, 174-175.	0.6	5
169	Clostridium PerfringensIntestinal Gas Gangrene in a Preterm Newborn. European Journal of Pediatric Surgery, 2009, 19, 257-259.	1.3	7
170	An Autophagic Vacuolar Myopathy-Like Disorder Presenting as Nonimmune Hydrops in a Female Fetus. Pediatric and Developmental Pathology, 2009, 12, 53-58.	1.0	6
171	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
172	Drug dosing error with dropsâ€"severe clinical course of codeine intoxication in twins. European Journal of Pediatrics, 2009, 168, 819-824.	2.7	39
173	Sudden unexpected death in an infant with L-2-hydroxyglutaric aciduria. European Journal of Pediatrics, 2009, 168, 957-962.	2.7	13
174	Inspiratory stridor and dysphagia in two newborn infants caused by ectopic thymus tissue. European Journal of Pediatrics, 2009, 168, 1141-1145.	2.7	15
175	Is serum procalcitonin a reliable diagnostic marker in children with acute respiratory tract infections? A retrospective analysis. European Journal of Pediatrics, 2009, 168, 1117-1124.	2.7	30
176	Development and implementation of a novel assay for <scp>l</scp>â€2â€hydroxyglutarate dehydrogenase (<scp>l</scp>â€2â€HGDH) in cell lysates: <scp>l</scp>â€2â€HGDH deficiency in 15 patients with <scp>l</scp>â€2â€hydroxyglutaric aciduria. Journal of Inherited Metabolic Disease, 2009, 32, 713-719.	3.6	8
177	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
178	Novel findings in two patients with lateâ€diagnosed afibrinogenaemia: intraosseous haemorrhage and fingertip necrosis. Haemophilia, 2009, 15, 980-982.	2.1	2
179	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
180	The Primordial Growth Disorder 3-M Syndrome Connects Ubiquitination to the Cytoskeletal Adaptor OBSL1. American Journal of Human Genetics, 2009, 84, 801-806.	6.2	93

#	Article	IF	Citations
181	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
182	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
183	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
184	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	6.2	81
185	Homozygous Inactivating Mutations in the NKX3-2 Gene Result in Spondylo-Megaepiphyseal-Metaphyseal Dysplasia. American Journal of Human Genetics, 2009, 85, 916-922.	6.2	30
186	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenatal Diagnosis, 2008, 28, 309-312.	2.3	45
187	Clinical and molecular analysis of arylsulfatase E in patients with brachytelephalangic chondrodysplasia punctata. American Journal of Medical Genetics, Part A, 2008, 146A, 997-1008.	1.2	36
188	Clinical and radiographic delineation of odontochondrodysplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 770-778.	1.2	23
189	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
190	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
191	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
192	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
193	Triangular tibia with fibular aplasia associated with a microdeletion on 2q11.2 encompassing LAF4. Clinical Genetics, 2008, 74, 560-565.	2.0	28
194	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
195	A novel mutation in the sulfate transporter gene SLC26A2 (DTDST) specific to the Finnish population causes de la Chapelle dysplasia. Journal of Medical Genetics, 2008, 45, 827-831.	3.2	22
196	Clinical and immunologic consequences of a somatic reversion in a patient with X-linked severe combined immunodeficiency. Blood, 2008, 112, 4090-4097.	1.4	59
197	Insights from a Transgenic Mouse Model on the Role of SLC26A2 in Health and Disease. Novartis Foundation Symposium, 2008, , 193-212.	1.1	4
198	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

#	Article	IF	Citations
199	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
200	Nosology and classification of genetic skeletal disorders: 2006 revision. American Journal of Medical Genetics, Part A, 2007, 143A, 1-18.	1.2	301
201	COL2A1â€“related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167.	1.2	32
202	Filamin A mutation is one cause of FG syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1876-1879.	1.2	44
203	A new case of spondyloenchondrodysplasia with immune dysregulation confirms the pleiotropic nature of the disorder: Comment on â€œA Syndrome of Immunodeficiency, Autoimmunity, and Spondylometaphyseal Dysplasiaâ€• by M.L. Kulkarni, K. Baskar, and P.M. Kulkarni [2006]. American Journal of Medical Genetics, Part A, 2007, 143A, 1394-1395.	1.2	7
204	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
205	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
206	Nucleotide-sugar transporter SLC35D1 is critical to chondroitin sulfate synthesis in cartilage and skeletal development in mouse and human. Nature Medicine, 2007, 13, 1363-1367.	30.7	103
207	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
208	L1CAM mutation in a boy with hydrocephalus and duplex kidneys. Pediatric Nephrology, 2007, 22, 1058-1061.	1.7	9
209	Early atherosclerosis in childhood type 1 diabetes: role of raised systolic blood pressure in the absence of dyslipidaemia. European Journal of Pediatrics, 2007, 166, 541-548.	2.7	45
210	Mutations in ACY1, the Gene Encoding Aminoacylase 1, Cause a Novel Inborn Error of Metabolism. American Journal of Human Genetics, 2006, 78, 401-409.	6.2	76
211	Torg Syndrome Is Caused by Inactivating Mutations in MMP2 and Is Allelic to NAO and Winchester Syndrome. Journal of Bone and Mineral Research, 2006, 22, 329-333.	2.8	63
212	Mutations in two regions ofFLNBresult in atelosteogenesis I and III. Human Mutation, 2006, 27, 705-710.	2.5	66
213	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
214	Spondylo-ocular syndrome: A new entity involving the eye and spine. American Journal of Medical Genetics, Part A, 2006, 140A, 652-656.	1.2	12
215	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	1.2	67
216	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity (Am J Med Genet 140A:) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.2	2

#	Article	IF	Citations
217	In vivo contribution of amino acid sulfur to cartilage proteoglycan sulfation. Biochemical Journal, 2006, 398, 509-514.	3.7	29
218	Deletion of Two Contiguous Genes, Platelet GPIbÎ² (Glycoprotein IbÎ²) and Septin SEPT5, in a Boy with Bernard-Soulier Syndrome and Developmental Delay: A Possible New Contiguous Gene Syndrome.. Blood, 2006, 108, 1097-1097.	1.4	0
219	Metaphyseal enchondrodysplasia with 2-hydroxy-glutaric aciduria: observation of a third case and further delineation. Clinical Dysmorphology, 2005, 14, 7-11.	0.3	15
220	Winchester syndrome caused by a homozygous mutation affecting the active site of matrix metalloproteinase 2. Clinical Genetics, 2005, 67, 261-266.	2.0	66
221	Identification of mutations in CUL7 in 3-M syndrome. Nature Genetics, 2005, 37, 1119-1124.	21.4	158
222	Dominant negative mutations in the C-propeptide of COL2A1 cause platyspondylic lethal skeletal dysplasia, torrance type, and define a novel subfamily within the type 2 collagenopathies. , 2005, 133A, 61-67.		56
223	Autosomal recessive omodysplasia: Early prenatal diagnosis and a possible clue to the gene location. American Journal of Medical Genetics, Part A, 2005, 135A, 324-327.	1.2	8
224	Mucolipidosis II presenting as severe neonatal hyperparathyroidism. European Journal of Pediatrics, 2005, 164, 236-243.	2.7	37
225	Familial X-linked cardiomyopathy (Danon disease): diagnostic confirmation by mutation analysis of the LAMP2gene. European Journal of Pediatrics, 2005, 164, 509-514.	2.7	43
226	Diagnosis of atelosteogenesis type II after a routine echography at 12 weeks' pregnancy. Prenatal Diagnosis, 2005, 25, 717-718.	2.3	5
227	The phenotypic spectrum in patients with arginine to cysteine mutations in the COL2A1 gene. Journal of Medical Genetics, 2005, 43, 406-413.	3.2	71
228	Evolutionary Comparison Provides Evidence for Pathogenicity of RMRP Mutations. PLoS Genetics, 2005, 1, e47.	3.5	57
229	Polymorphisms and Haplotypes of Acid Mammalian Chitinase Are Associated with Bronchial Asthma. American Journal of Respiratory and Critical Care Medicine, 2005, 172, 1505-1509.	5.6	129
230	A diastrophic dysplasia sulfate transporter (SLC26A2) mutant mouse: morphological and biochemical characterization of the resulting chondrodysplasia phenotype. Human Molecular Genetics, 2005, 14, 859-871.	2.9	116
231	Morphological, clinical and genetic aspects in a family with a novel LAMP-2 gene mutation (Danon) Tj ETQq1 1 0.784314 rgBT /Overlo	0.6	30
232	Metaphyseal enchondrodysplasia with 2-hydroxy-glutaric aciduria: observation of a third case and further delineation. Clinical Dysmorphology, 2005, 14, 7-11.	0.3	6
233	Growing bone knowledge. Clinical Genetics, 2004, 66, 399-401.	2.0	5
234	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252

#	Article	IF	Citations
235	Clinical, radiographic, and genetic diagnosis of progressive pseudorheumatoid dysplasia in a patient with severe polyarthropathy. Rheumatology International, 2004, 24, 53-56.	3.0	49
236	Spondyloperipheral dysplasia is caused by truncating mutations in the C-propeptide of COL2A1. , 2004, 129A, 144-148.		36
237	Phenotypic and molecular characterization of Bruck syndrome (osteogenesis imperfecta with) Tj ETQq1 1 0.784314 rgBT /Overlock 1 Medical Genetics, Part A, 2004, 131A, 115-120.	1.2	146
238	Novel mutation in the tyrosine kinase domain of FGFR2 in a patient with Pfeiffer syndrome. , 2004, 131A, 299-300.		16
239	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
240	An 11-month-old boy with psychomotor regression and auto-aggressive behaviour. European Journal of Pediatrics, 2003, 162, 559-561.	2.7	20
241	Leigh syndrome due to compound heterozygosity of dihydrolipoamide dehydrogenase gene mutations. Description of the first E3 splice site mutation. European Journal of Pediatrics, 2003, 162, 714-718.	2.7	53
242	Glutaric aciduria type 1 and neonatal screening: time to proceed?with caution. European Journal of Pediatrics, 2003, 162, S17-S20.	2.7	8
243	Prenatal diagnosis of boomerang dysplasia. , 2003, 122A, 148-154.		13
244	Autosomal recessive multiple epiphyseal dysplasia with homozygosity for C653S in theDTDSTgene: Double-layer patella as a reliable sign. , 2003, 122A, 187-192.		53
245	A cluster of autosomal recessive spondylocostal dysostosis caused by three newly identified DLL3 mutations segregating in a small village. Clinical Genetics, 2003, 64, 28-35.	2.0	38
246	Mutations in ENPP1 are associated with 'idiopathic' infantile arterial calcification. Nature Genetics, 2003, 34, 379-381.	21.4	539
247	Mutations in the Gene Encoding Capillary Morphogenesis Protein 2 Cause Juvenile Hyaline Fibromatosis and Infantile Systemic Hyalinosis. American Journal of Human Genetics, 2003, 73, 791-800.	6.2	209
248	Recessive multiple epiphyseal dysplasia (rMED): phenotype delineation in eighteen homozygotes for DTDST mutation R279W. Journal of Medical Genetics, 2003, 40, 65-71.	3.2	67
249	Osteoclast Morphology in Autosomal Recessive Malignant Osteopetrosis Due To a TCIRG1 Gene Mutation. Fetal and Pediatric Pathology, 2003, 22, 3-9.	0.3	8
250	A recurrent R718W mutation in COMP results in multiple epiphyseal dysplasia with mild myopathy: clinical and pathogenetic overlap with collagen IX mutations. Journal of Medical Genetics, 2003, 40, 942-948.	3.2	26
251	A Diagnostic Approach to Skeletal Dysplasias. , 2003, , 375-IX.		7
252	Osteoclast Morphology in Autosomal Recessive Malignant Osteopetrosis Due To a TCIRG1 Gene Mutation. Fetal and Pediatric Pathology, 2003, 22, 3-9.	0.3	2

#	Article	IF	Citations
253	DTDST mutations are not a frequent cause of idiopathic talipes equinovarus (club foot). Journal of Medical Genetics, 2002, 39, 20e-20.	3.2	8
254	Quality of life and psychologic adjustment in children and adolescents with early treated phenylketonuria can be normal. Journal of Pediatrics, 2002, 140, 516-521.	1.8	93
255	The Gene for Juvenile Hyaline Fibromatosis Maps to Chromosome 4q21. American Journal of Human Genetics, 2002, 71, 975-980.	6.2	71
256	Analysis of the CTNS gene in patients of German and Swiss origin with nephropathic cystinosis. Human Mutation, 2002, 20, 237-237.	2.5	32
257	Hyperammonaemic encephalopathy in a 13-year-old boy. European Journal of Pediatrics, 2002, 161, 163-164.	2.7	5
258	RMRP gene sequence analysis confirms a cartilage-hair hypoplasia variant with only skeletal manifestations and reveals a high density of single-nucleotide polymorphisms. Clinical Genetics, 2002, 61, 146-151.	2.0	77
259	PC-1 Nucleoside Triphosphate Pyrophosphohydrolase Deficiency in Idiopathic Infantile Arterial Calcification. American Journal of Pathology, 2001, 158, 543-554.	3.8	275
260	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
261	Hepatic Carnitine Palmitoyltransferase I Deficiency: Acylcarnitine Profiles in Blood Spots Are Highly Specific. Clinical Chemistry, 2001, 47, 1763-1768.	3.2	110
262	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
263	Novel missense mutations outside the allosteric domain of glutamate dehydrogenase are prevalent in European patients with the congenital hyperinsulinism-hyperammonemia syndrome. Human Genetics, 2001, 108, 66-71.	3.8	51
264	Mutations in the diastrophic dysplasia sulfate transporter (DTDST) gene (SLC26A2): 22 novel mutations, mutation review, associated skeletal phenotypes, and diagnostic relevance. Human Mutation, 2001, 17, 159-171.	2.5	173
265	Molecular-pathogenetic classification of genetic disorders of the skeleton. American Journal of Medical Genetics Part A, 2001, 106, 282-293.	2.4	126
266	The mutational spectrum of human malignant autosomal recessive osteopetrosis. Human Molecular Genetics, 2001, 10, 1767-1773.	2.9	201
267	Boy with syndactylies, macrocephaly, and severe skeletal dysplasia: Not a new syndrome, but two dominant mutations (GLI3 E543X andCOL2A1 G973R) in the same individual. American Journal of Medical Genetics Part A, 2000, 90, 239-242.	2.4	14
268	Otopalatodigital syndrome and frontometaphyseal dysplasia, splitters and lumpers, and paternity of ideas. American Journal of Medical Genetics Part A, 2000, 95, 86-86.	2.4	5
269	A 17-month-old boy with bowed legs. European Journal of Pediatrics, 2000, 159, 863-865.	2.7	1
270	Clinical variability of Stickler syndrome with a COL2A1 haploinsufficiency mutation: implications for genetic counselling. Journal of Medical Genetics, 2000, 37, 318-320.	3.2	22

#	Article	IF	Citations
271	Autosomal Recessive Disorder Otospondylomegaepiphyseal Dysplasia Is Associated with Loss-of-Function Mutations in the COL11A2 Gene. American Journal of Human Genetics, 2000, 66, 368-377.	6.2	78
272	Clinical and Genetic Features of Ehlersâ€“Danlos Syndrome Type IV, the Vascular Type. New England Journal of Medicine, 2000, 342, 673-680.	27.0	1,219
273	Achondroplasia with the FGFR3 1138gright-arrowa (G380R) mutation in two sibs sharing a 4p haplotype derived from their unaffected father. Journal of Medical Genetics, 2000, 37, 958-959.	3.2	22
274	Point Mutations Throughout the GLI3 Gene Cause Greig Cephalopolysyndactyly Syndrome. Human Molecular Genetics, 1999, 8, 1769-1777.	2.9	131
275	Homozygosity for a novel DTDST mutation in a child with a â€˜broad bone-platyspondylicâ€™ variant of diastrophic dysplasia. Clinical Genetics, 1999, 56, 71-76.	2.0	16
276	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	21.4	260
277	Determination of Bone Markers in Pycnodysostosis: Effects of Cathepsin K Deficiency on Bone Matrix Degradation. Journal of Bone and Mineral Research, 1999, 14, 1902-1908.	2.8	128
278	The painful hip: evaluation of criteria for clinical decision-making. European Journal of Pediatrics, 1999, 158, 923-928.	2.7	93
279	Ehlers-Danlos Syndrome Type VII. Journal of Bone and Joint Surgery - Series A, 1999, 81, 225-238.	3.0	74
280	Diagnosis and management of glutaric aciduria type I. Journal of Inherited Metabolic Disease, 1998, 21, 326-340.	3.6	105
281	Mutations in orthologous genes in human spondyloepimetaphyseal dysplasia and the brachymorphic mouse. Nature Genetics, 1998, 20, 157-162.	21.4	189
282	Identification of fifteen novel mutations in the tissue-nonspecific alkaline phosphatase (TNSALP) gene in European patients with severe hypophosphatasia. European Journal of Human Genetics, 1998, 6, 308-314.	2.8	122
283	Mutation and deletion of the pseudoautosomal gene SHOX cause Leri-Weill dyschondrosteosis. Nature Genetics, 1998, 19, 70-73.	21.4	316
284	Schwartz-Jampel syndrome type 2 and Stï¿½ve-Wiedemann syndrome: A case for ?Lumping?. , 1998, 78, 150-154.		43
285	Prenatal ultrasonographic description and postnatal pathological findings in atelosteogenesis type 1. , 1998, 79, 392-395.		19
286	The human glutaryl-CoA dehydrogenase gene: report of intronic sequences and of 13 novel mutations causing glutaric aciduria type I. Human Genetics, 1998, 102, 452-458.	3.8	54
287	Proteoglycan sulfation in cartilage and cell cultures from patients with sulfate transporter chondrodysplasias: Relationship to clinical severity and indications on the role of intracellular sulfate production. Matrix Biology, 1998, 17, 361-369.	3.6	59
288	The Effect of the N-methyl-D-aspartate Receptor Antagonist Dextromethorphan on Perioperative Brain Injury in Children Undergoing Cardiac Surgery with Cardiopulmonary Bypass: Results of a Pilot Study. Neuropediatrics, 1997, 28, 191-197.	0.6	21

#	Article	IF	Citations
289	Undersulfation of Cartilage Proteoglycans Ex Vivo and Increased Contribution of Amino Acid Sulfur to Sulfation In Vitro in McAlister Dysplasia/Atelosteogenesis Type 2. FEBS Journal, 1997, 248, 741-747.	0.2	29
290	Heterogeneity in Schwartz-Jampel chondrodystrophic myotonia. European Journal of Pediatrics, 1997, 156, 214-223.	2.7	69
291	Detection and characterization of mitochondrial DNA rearrangements in Pearson and Kearns-Sayre syndromes by long PCR. Human Genetics, 1997, 100, 643-650.	3.8	52
292	Sulfate Transport in Chondrodysplasia,a. Annals of the New York Academy of Sciences, 1996, 785, 131-136.	3.8	17
293	A Family of Chondrodysplasias Caused by Mutations in the Diastrophic Dysplasia Sulfate Transporter Gene and Associated with Impaired Sulfation of Proteoglycansa. Annals of the New York Academy of Sciences, 1996, 785, 195-201.	3.8	24
294	Phenotypic and genotypic overlap between atelosteogenesis type 2 and diastrophic dysplasia. Human Genetics, 1996, 98, 657-661.	3.8	54
295	A specific collagen type II gene (COL2A1) mutation presenting as spondyloperipheral dysplasia. , 1996, 63, 123-128.		45
296	A chondrodysplasia family produced by mutations in thediastrophic dysplasia sulfate transporter gene: Genotype/phenotype correlations. American Journal of Medical Genetics Part A, 1996, 63, 144-147.	2.4	114
297	Achondrogenesis type IB is caused by mutations in the diastrophic dysplasia sulphate transporter gene. Nature Genetics, 1996, 12, 100-102.	21.4	219
298	The deletion of six amino acids at the C-terminus of the alpha 1 (II) chain causes overmodification of type II and type XI collagen: further evidence for the association between small deletions in COL2A1 and Kniest dysplasia.. Journal of Medical Genetics, 1996, 33, 649-654.	3.2	20
299	Undersulfation of Proteoglycans Synthesized by Chondrocytes from a Patient with Achondrogenesis Type 1B Homozygous for an L483P Substitution in the Diastrophic Dysplasia Sulfate Transporter. Journal of Biological Chemistry, 1996, 271, 18456-18464.	3.4	68
300	Achondrogenesis type 1B.. Journal of Medical Genetics, 1996, 33, 957-961.	3.2	28
301	Clinical Course, Early Diagnosis, Treatment, and Prevention of Disease in Glutaryl-CoA Dehydrogenase Deficiency. Neuropediatrics, 1996, 27, 115-123.	0.6	237
302	Ehlersâ€Danlos syndrome type IV caused by Gly400Glu, Gly595Cys and Glyl003Asp substitutions in collagen III: clinical features, biochemical screening, and molecular confirmation. Clinical Genetics, 1996, 49, 286-295.	2.0	7
303	Phenotype of the Williams-Beuren syndrome associated with hemizygosity at the elastin locus. European Journal of Pediatrics, 1995, 154, 477-482.	2.7	48
304	Early signs and course of disease of glutaryl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1995, 18, 173-176.	3.6	19
305	Feline Mucopolysaccharidosis VII Due to Î²-glucuronidase Deficiency. Veterinary Pathology, 1994, 31, 435-443.	1.7	66
306	Prenatal Diagnosis of Collagen Disorders by Direct Biochemical Analysis of Chorionic Villus Biopsies. Pediatric Research, 1994, 36, 441-448.	2.3	51

#	Article	IF	Citations
307	Somatosensory Evoked Potentials with High Cortical Amplitudes: Clinical Data in 31 Children. Neuropediatrics, 1994, 25, 78-84.	0.6	18
308	Decreased extracellular deposition of fibrillin and decorin in neonatal Marfan syndrome fibroblasts. Human Genetics, 1993, 90, 511-5.	3.8	56
309	Pearson bone marrow-pancreas syndrome with insulin-dependent diabetes, progressive renal tubulopathy, organic aciduria and elevated fetal haemoglobin caused by deletion and duplication of mitochondrial DNA. European Journal of Pediatrics, 1993, 152, 44-50.	2.7	74
310	Complementary DNA Sequence and Chromosomal Mapping of a Human Proteoglycan-Binding Cell-Adhesion Protein (Dermatopontin). Genomics, 1993, 17, 463-467.	2.9	50
311	An Intronic Deletion Leading to Skipping of Exon 21 ofCol1a2in a Boy with Mild Osteogenesis Imperfecta. Connective Tissue Research, 1993, 29, 31-40.	2.3	11
312	Hypercalciuria and Nephrocalcinosis, a Feature of Wilsonâ€™s Disease. Nephron, 1993, 65, 460-462.	1.8	42
313	Deficiencies of fibrillin and decorin in fibroblast cultures of a patient with neonatal Marfan syndrome.. Journal of Medical Genetics, 1992, 29, 875-878.	3.2	22
314	Mechanism of action of FK 506 and cyclosporin. Lancet, The, 1991, 337, 439.	13.7	7
315	Maternal phenylketonuria syndrome in cousins caused by mild, unrecognized phenylketonuria in their mothers homozygous for the phenylalanine hydroxylase Arg-261-Gln mutation. European Journal of Pediatrics, 1991, 150, 493-497.	2.7	15
316	Radiological â€œmetamorphosisâ€•in a patient with severe congenital osteogenesis imperfecta. European Journal of Pediatrics, 1990, 149, 403-405.	2.7	3
317	AHaeIII RFLP in COL1A1. Nucleic Acids Research, 1990, 18, 5926-5926.	14.5	3
318	Characterization of Large Deletions in the Pro-?1(III) mRNA from Two Ehlers-Danlos Type IV Patients. Annals of the New York Academy of Sciences, 1990, 580, 552-553.	3.8	0
319	Clinical variability of osteogenesis imperfecta linked to COL1A2 and associated with a structural defect in the type I collagen molecule.. Journal of Medical Genetics, 1989, 26, 358-362.	3.2	28
320	Ehlers-Danlos syndrome type IV: A subset of patients distinguished by low serum levels of the amino-terminal propeptide of type III procollagen. American Journal of Medical Genetics Part A, 1989, 34, 68-71.	2.4	28
321	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. Human Genetics, 1989, 82, 104-108.	3.8	97
322	TYPE m COLLAGEN DEFICIENCY. Lancet, The, 1989, 333, 903-904.	13.7	12
323	Ehlers-Danlos syndrome type IV: Another temperature-dependent skin disorder?. Journal of the American Academy of Dermatology, 1989, 21, 323.	1.2	1
324	?Cerebral? lactic acidosis and cerebrospinal fluid pH. European Journal of Pediatrics, 1988, 147, 667-668.	2.7	1

#	Article	IF	Citations
325	Impaired secretion of type III procollagen in Ehlers-Danlos syndrome type IV fibroblasts: Correction of the defect by incubation at reduced temperature and demonstration of subtle alterations in the triple-helical region of the molecule. Biochemical and Biophysical Research Communications, 1988, 150, 140-147.	2.1	42
326	Imperfect Collagenesis in Osteogenesis Imperfecta The Consequences of Cysteine-Glycine Substitutions upon Collagen Structure and Metabolism. Annals of the New York Academy of Sciences, 1988, 543, 47-61.	3.8	15
327	Normal Thermal Stability of an Overmodified Type I Collagen Despite a Structural Mutation within the Triple Helical Region in a Case of Osteogenesis Irnperfecta Type IVB. Annals of the New York Academy of Sciences, 1988, 543, 83-84.	3.8	2
328	Delayed Triple-Helix Formation of Abnormal Type I Collagen Is Corrected by Reduced Temperature.. Annals of the New York Academy of Sciences, 1988, 543, 85-92.	3.8	4
329	CONGENITAL DOPAMINE BETA-HYDROXYLASE DEFICIENCY. Lancet, The, 1987, 329, 693.	13.7	7
330	Galactosemia caused by generalized uridine diphosphate galactose-4-epimerase deficiency. Journal of Pediatrics, 1986, 109, 1074-1075.	1.8	4
331	Galactosemia caused by generalized uridine diphosphate galactose-4-epimerase deficiency. Journal of Pediatrics, 1983, 103, 927-930.	1.8	19
332	Skeletal Dysplasias Related to Defects in Sulfate Metabolism. , 0, , 939-960.		15
333	The Ehlers-Danlos Syndrome. , 0, , 431-523.		175
334	A Novel Talin-to-RhoA Switch Mechanism Upon Ligand Binding of the Collagen VI Receptor CMG2. SSRN Electronic Journal, 0, , .	0.4	1