Andrea Superti-Furga

List of Publications by Year
in descending order

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334 papers	20,530 citations	15880 67 h-index	16186 128 g-index
351 all docs	351 docs citations	351 times ranked	21296 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.	0.7	3
2	Clinical and Molecular Diagnosis of Osteocraniostenosis in Fetuses and Newborns: Prenatal Ultrasound, Clinical, Radiological and Pathological Features. Genes, 2022, 13, 261.	1.0	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.	2.6	29
4	SCN5A Overlap Syndromes: an open-minded approach. Heart Rhythm, 2022, , .	0.3	2
5	Clinical and Genetic Findings in a Series of Eight Families with Arthrogryposis. Genes, 2022, 13, 29.	1.0	6
6	Identification of Disease Gene for Camurati-Engelmann Disease, Type II. Bone Reports, 2022, 16, 101561.	0.2	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.	1.5	5
8	Cancer surveillance in children with Ollier Disease and Maffucci Syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1338-1340.	0.7	2
9	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	13.7	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.	1.4	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.	1.0	4
12	Spinal cerebrotendinous xanthomatosis: A case report and literature review. Molecular Genetics and Metabolism Reports, 2021, 26, 100719.	0.4	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.	2.0	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.	0.7	3
15	NGS-Based Diagnosis of Treatable Neurogenetic Disorders in Adults: Opportunities and Challenges. Genes, 2021, 12, 695.	1.0	5
16	Case Report: A Rare Truncating Variant of the CFHR5 Gene in IgA Nephropathy. Frontiers in Genetics, 2021, 12, 529236.	1.1	3
17	Classical homocystinuria, is it safe to exercise?. Molecular Genetics and Metabolism Reports, 2021, 27, 100746.	0.4	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.	1.1	3

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19	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	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.	1.5	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.	0.7	0
22	Elevated lactate in Mauriac syndrome: still a mystery. BMC Endocrine Disorders, 2021, 21, 172.	0.9	1
23	CNV Detection from Exome Sequencing Data in Routine Diagnostics of Rare Genetic Disorders: Opportunities and Limitations. Genes, 2021, 12, 1427.	1.0	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.	1.0	1
25	Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. Scientific Reports, 2021, 11, 19332.	1.6	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.	0.4	7
27	AutoMap is a high performance homozygosity mapping tool using next-generation sequencing data. Nature Communications, 2021, 12, 518.	5.8	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.	0.7	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.	2.6	12
30	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	1.1	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.	1.0	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.	3.1	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.	1.2	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.	1.4	14
35	Non-invasive prenatal testing leading to a maternal diagnosis of Charcotâ€“Marieâ€“Tooth neuropathy. Journal of Human Genetics, 2020, 65, 1035-1038.	1.1	4
36	Clinical aspects of Hyaline Fibromatosis Syndrome and identification of a novel mutation. Molecular Genetics & Genomic Medicine, 2020, 8, e1203.	0.6	13

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37	Childhood neurodegeneration associated with a specific UBTF variant: a new case report and review of the literature. BMC Neurology, 2020, 20, 17.	0.8	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.	1.0	9
39	Skeletal Dysplasias Caused by Sulfation Defects. International Journal of Molecular Sciences, 2020, 21, 2710.	1.8	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.	1.3	1
42	Bone and connective tissue disorders caused by defects in glycosaminoglycan biosynthesis: a panoramic view. FEBS Journal, 2019, 286, 3008-3032.	2.2	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.	1.1	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.	1.0	13
45	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	0.7	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.	1.7	15
47	A novel missense variant in IDH3A causes autosomal recessive retinitis pigmentosa. Ophthalmic Genetics, 2019, 40, 177-181.	0.5	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.	0.1	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.	2.6	17
51	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	1.4	11
52	Progressive pseudorheumatoid dysplasia: a rare childhood disease. Rheumatology International, 2019, 39, 441-452.	1.5	22
53	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. JCI Insight, 2019, 4, .	2.3	30
54	Dysostosen. Springer Reference Medizin, 2019, , 1-12.	0.0	0

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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.	0.1	0
57	Confirmation of spondyloâ€epiâ€metaphyseal dysplasia with joint laxity, EXOC6B type. American Journal of Medical Genetics, Part A, 2018, 176, 2934-2935.	0.7	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	0.1	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.	0.7	10
60	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	4.2	76
61	Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1739-1746.	0.7	24
62	The multiple faces of artwork diagnoses. Lancet Neurology, The, 2017, 16, 417.	4.9	4
63	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. Nature Communications, 2017, 8, 15861.	5.8	56
64	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	2.6	90
65	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	0.7	38
66	Genetic disorders of bone â€“ An historical perspective. Bone, 2017, 102, 1-4.	1.4	5
67	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€. American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
68	Chondroitin SulfateN-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. Human Mutation, 2017, 38, 34-38.	1.1	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.	1.1	94
71	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	0.7	8
72	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254

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73	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	9.4	125
74	Natural history and life-threatening complications in Myhre syndrome and review of the literature. European Journal of Pediatrics, 2016, 175, 1307-1315.	1.3	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.	2.6	52
76	Bone Formation and the Wnt Signaling Pathway. New England Journal of Medicine, 2016, 375, 1902-1903.	13.9	19
77	Bisphosphonates in multicentric osteolysis, nodulosis and arthropathy (MONA) spectrum disorder â€“ an alternative therapeutic approach. Scientific Reports, 2016, 6, 34017.	1.6	20
78	BGN Mutations in X-Linked Spondyloepimetaphyseal Dysplasia. American Journal of Human Genetics, 2016, 98, 1243-1248.	2.6	29
79	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	13.9	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.	0.7	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.	2.9	17
82	Mutations in LONP1, a mitochondrial matrix protease, cause CODAS syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1501-1509.	0.7	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.	1.6	65
84	Analysis of the genetic basis of periodic fever with aphthous stomatitis, pharyngitis and cervical adenitis (PFAPA) syndrome. Scientific Reports, 2015, 5, 10200.	1.6	70
85	Significant clinical benefits of molecular studies in the skeletal dysplasias. American Journal of Medical Genetics, Part A, 2015, 167, 476-477.	0.7	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.	0.7	66
87	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	0.7	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.	0.7	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.	3.1	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.	1.4	70

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91	N-acetylcysteine treatment ameliorates the skeletal phenotype of a mouse model of diastrophic dysplasia. Human Molecular Genetics, 2015, 24, 5570-5580.	1.4	22
92	Multiple sulfatase deficiency with neonatal manifestation. Italian Journal of Pediatrics, 2014, 40, 86.	1.0	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.	0.7	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.	0.7	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.	3.3	56
97	Acampomelic Form of Campomelic Dysplasia with SOX9 Missense Mutation. Indian Journal of Pediatrics, 2014, 81, 98-100.	0.3	8
98	Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone, 2014, 59, 122-126.	1.4	26
99	MMP13 mutations are the cause of recessive metaphyseal dysplasia, Spahr type. American Journal of Medical Genetics, Part A, 2014, 164, 1175-1179.	0.7	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.	1.2	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.	3.3	73
103	3-M syndrome associated with growth hormone deficiency: 18Âyear follow-up of a patient. Italian Journal of Pediatrics, 2013, 39, 21.	1.0	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.	0.7	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.	0.7	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.	1.1	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.	2.6	112
108	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	2.6	114

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109	Exome Sequencing Identifies INPPL1 Mutations as a Cause of Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 144-149.	2.6	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.	0.7	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.	1.5	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.	0.7	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.	0.7	17
114	The dark sides of capillary morphogenesis gene 2. EMBO Journal, 2012, 31, 3-13.	3.5	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.	0.7	2
116	Metaphyseal chondromatosis combined with D-2-hydroxyglutaric aciduria in four patients. Skeletal Radiology, 2012, 41, 1479-1487.	1.2	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.	1.1	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.	0.7	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.	0.7	12
121	Enchondromatosis revisited: New classification with molecular basis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 154-164.	0.7	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.	0.7	74
123	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	0.7	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.	2.6	192

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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.	2.6	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.	1.1	104
129	Mutation analysis in 54 propionic acidemia patients. Journal of Inherited Metabolic Disease, 2012, 35, 51-63.	1.7	41
130	Propionic acidemia: neonatal versus selective metabolic screening. Journal of Inherited Metabolic Disease, 2012, 35, 41-49.	1.7	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.	1.5	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.	1.8	37
134	Clinical and molecular characterization of Diastrophic Dysplasia in the Portuguese population. Clinical Genetics, 2011, 80, 550-557.	1.0	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.	1.9	29
136	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	9.4	151
137	Hyperpyrexia resulting in encephalopathy in a 14-month-old patient with cblC disease. Brain and Development, 2011, 33, 432-436.	0.6	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.	2.6	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.	2.6	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.	2.6	31
141	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. Human Genetics, 2011, 129, 497-502.	1.8	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.	0.7	19
143	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	0.7	573
144	Axial spondylometaphyseal dysplasia: Additional reports. American Journal of Medical Genetics, Part A, 2011, 155, 2521-2528.	0.7	8

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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.	0.7	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.	0.7	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.	0.7	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.	3.3	45
149	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	3.1	149
150	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	1.5	39
151	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	1.5	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.	2.6	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.	0.8	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.	0.7	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.	0.7	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.	0.7	67
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