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	LDL Receptor-Related Protein 5 (LRP5) Affects Bone Accrual and Eye Development. Cell, 2001, 107, 513-523.	28.9	2,055
2	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
3	Nosology and classification of genetic skeletal disorders: 2010 revision. American Journal of Medical Genetics, Part A, 2011, 155, 943-968.	1.2	573
4	Mutations in ENPP1 are associated with 'idiopathic' infantile arterial calcification. Nature Genetics, 2003, 34, 379-381.	21.4	539
5	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	1.2	453
6	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
7	Mutation and deletion of the pseudoautosomal gene SHOX cause Leri-Weill dyschondrosteosis. Nature Genetics, 1998, 19, 70-73.	21.4	316
8	Nosology and classification of genetic skeletal disorders: 2006 revision. American Journal of Medical Genetics, Part A, 2007, 143A, 1-18.	1.2	301
9	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
10	PC-1 Nucleoside Triphosphate Pyrophosphohydrolase Deficiency in Idiopathic Infantile Arterial Calcification. American Journal of Pathology, 2001, 158, 543-554.	3.8	275
11	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	21.4	260
12	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
13	Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 2004, 36, 405-410.	21.4	252
14	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
15	Clinical Course, Early Diagnosis, Treatment, and Prevention of Disease in Glutaryl-CoA Dehydrogenase Deficiency. Neuropediatrics, 1996, 27, 115-123.	0.6	237
16	Achondrogenesis type IB is caused by mutations in the diastrophic dysplasia sulphate transporter gene. Nature Genetics, 1996, 12, 100-102.	21.4	219
17	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
18	The mutational spectrum of human malignant autosomal recessive osteopetrosis. Human Molecular Genetics, 2001, 10, 1767-1773.	2.9	201

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19	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
20	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
21	Mutations in orthologous genes in human spondyloepimetaphyseal dysplasia and the brachymorphic mouse. Nature Genetics, 1998, 20, 157-162.	21.4	189
22	The Ehlers-Danlos Syndrome. , 0, , 431-523.		175
23	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
24	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
25	Identification of mutations in CUL7 in 3-M syndrome. Nature Genetics, 2005, 37, 1119-1124.	21.4	158
26	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	21.4	151
27	Mutations in FKBP10 cause recessive osteogenesis imperfecta and bruck syndrome. Journal of Bone and Mineral Research, 2011, 26, 666-672.	2.8	149
28	Phenotypic and molecular characterization of Bruck syndrome (osteogenesis imperfecta with) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 387 Medical Genetics, Part A, 2004, 131A, 115-120.	1.2	146
29	Propionic acidemia: clinical course and outcome in 55 pediatric and adolescent patients. Orphanet Journal of Rare Diseases, 2013, 8, 6.	2.7	138
30	Point Mutations Throughout the GLI3 Gene Cause Greig Cephalopolysyndactyly Syndrome. Human Molecular Genetics, 1999, 8, 1769-1777.	2.9	131
31	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
32	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
33	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
34	Molecular-pathogenetic classification of genetic disorders of the skeleton. American Journal of Medical Genetics Part A, 2001, 106, 282-293.	2.4	126
35	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. Nature Genetics, 2016, 48, 777-784.	21.4	125
36	Hyperpyrexia resulting in encephalopathy in a 14-month-old patient with cblC disease. Brain and Development, 2011, 33, 432-436.	1.1	123

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37	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
38	Lethal Skeletal Dysplasia in Mice and Humans Lacking the Golgin GMAP-210. New England Journal of Medicine, 2010, 362, 206-216.	27.0	122
39	Cortical-Bone Fragility â€" Insights from sFRP4 Deficiency in Pyleâ€™s Disease. New England Journal of Medicine, 2016, 374, 2553-2562.	27.0	119
40	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
41	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
42	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	6.2	114
43	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
44	Hepatic Carnitine Palmitoyltransferase I Deficiency: Acylcarnitine Profiles in Blood Spots Are Highly Specific. Clinical Chemistry, 2001, 47, 1763-1768.	3.2	110
45	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
46	Diagnosis and management of glutaric aciduria type I. Journal of Inherited Metabolic Disease, 1998, 21, 326-340.	3.6	105
47	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	3.5	104
48	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
49	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
50	Molecular defects of type III procollagen in Ehlers-Danlos syndrome type IV. Human Genetics, 1989, 82, 104-108.	3.8	97
51	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
52	The painful hip: evaluation of criteria for clinical decision-making. European Journal of Pediatrics, 1999, 158, 923-928.	2.7	93
53	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
54	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

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55	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
56	DOMINO: Using Machine Learning to Predict Genes Associated with Dominant Disorders. American Journal of Human Genetics, 2017, 101, 623-629.	6.2	90
57	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
58	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	6.2	81
59	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
60	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
61	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
62	EXTL3 mutations cause skeletal dysplasia, immune deficiency, and developmental delay. Journal of Experimental Medicine, 2017, 214, 623-637.	8.5	76
63	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
64	Prostaglandin transporter mutations cause pachydermoperiostosis with myelofibrosis. Human Mutation, 2012, 33, 1175-1181.	2.5	74
65	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
66	Ehlers-Danlos Syndrome Type VII. Journal of Bone and Joint Surgery - Series A, 1999, 81, 225-238.	3.0	74
67	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
68	The Gene for Juvenile Hyaline Fibromatosis Maps to Chromosome 4q21. American Journal of Human Genetics, 2002, 71, 975-980.	6.2	71
69	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
70	The dark sides of capillary morphogenesis gene 2. EMBO Journal, 2012, 31, 3-13.	7.8	71
71	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	1.6	71
72	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

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73	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
74	Heterogeneity in Schwartz-Jampel chondrodystrophic myotonia. European Journal of Pediatrics, 1997, 156, 214-223.	2.7	69
75	Propionic acidemia: neonatal versus selective metabolic screening. Journal of Inherited Metabolic Disease, 2012, 35, 41-49.	3.6	69
76	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
77	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. Human Genetics, 2011, 129, 497-502.	3.8	68
78	AutoMap is a high performance homozygosity mapping tool using next-generation sequencing data. Nature Communications, 2021, 12, 518.	12.8	68
79	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
80	Frontometaphyseal dysplasia: Mutations inFLNA and phenotypic diversity. American Journal of Medical Genetics, Part A, 2006, 140A, 1726-1736.	1.2	67
81	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
82	Feline Mucopolysaccharidosis VII Due to Î²-glucuronidase Deficiency. Veterinary Pathology, 1994, 31, 435-443.	1.7	66
83	Winchester syndrome caused by a homozygous mutation affecting the active site of matrix metalloproteinase 2. Clinical Genetics, 2005, 67, 261-266.	2.0	66
84	Mutations in two regions ofFLNBresult in atelosteogenesis I and III. Human Mutation, 2006, 27, 705-710.	2.5	66
85	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
86	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
87	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
88	Mutations in LONP1, a mitochondrial matrix protease, cause CODAS syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1501-1509.	1.2	61
89	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
90	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

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91	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
92	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
93	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
94	Evolutionary Comparison Provides Evidence for Pathogenicity of RMRP Mutations. PLoS Genetics, 2005, 1, e47.	3.5	57
95	Decreased extracellular deposition of fibrillin and decorin in neonatal Marfan syndrome fibroblasts. Human Genetics, 1993, 90, 511-5.	3.8	56
96	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
97	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
98	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
99	Molecular pathogenesis of Spondylocheirodysplastic Ehlersâ€Ðanlos syndrome caused by mutant ZIP13 proteins. EMBO Molecular Medicine, 2014, 6, 1028-1042.	6.9	56
100	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. Nature Communications, 2017, 8, 15861.	12.8	56
101	Phenotypic and genotypic overlap between atelosteogenesis type 2 and diastrophic dysplasia. Human Genetics, 1996, 98, 657-661.	3.8	54
102	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
103	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
104	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
105	Autosomal recessive multiple epiphyseal dysplasia with homozygosity for C653S in theDTDSTgene: Double-layer patella as a reliable sign. , 2003, 122A, 187-192.		53
106	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
107	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
108	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

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109	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
110	Prenatal Diagnosis of Collagen Disorders by Direct Biochemical Analysis of Chorionic Villus Biopsies. Pediatric Research, 1994, 36, 441-448.	2.3	51
111	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
112	Complementary DNA Sequence and Chromosomal Mapping of a Human Proteoglycan-Binding Cell-Adhesion Protein (Dermatopontin). Genomics, 1993, 17, 463-467.	2.9	50
113	Clinical, radiographic, and genetic diagnosis of progressive pseudorheumatoid dysplasia in a patient with severe polyarthropathy. Rheumatology International, 2004, 24, 53-56.	3.0	49
114	Phenotype of the Williams-Beuren syndrome associated with hemizygosity at the elastin locus. European Journal of Pediatrics, 1995, 154, 477-482.	2.7	48
115	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
116	A specific collagen type II gene (COL2A1) mutation presenting as spondyloperipheral dysplasia. , 1996, 63, 123-128.		45
117	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
118	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenatal Diagnosis, 2008, 28, 309-312.	2.3	45
119	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
120	TRPV4-pathy, a novel channelopathy affecting diverse systems. Journal of Human Genetics, 2010, 55, 400-402.	2.3	45
121	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
122	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
123	Filamin A mutation is one cause of FG syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 1876-1879.	1.2	44
124	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
125	Exome Sequencing Identifies INPPL1 Mutations as a Cause of Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 144-149.	6.2	44
126	Schwartz-Jampel syndrome type 2 and Stï¿½ve-Wiedemann syndrome: A case for ?Lumping?. , 1998, 78, 150-154.		43

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127	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
128	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
129	Hypercalciuria and Nephrocalcinosis, a Feature of Wilsonâ€™s Disease. Nephron, 1993, 65, 460-462.	1.8	42
130	Bone Dysplasias. , 2012, , .		42
131	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
132	Mutation analysis in 54 propionic acidemia patients. Journal of Inherited Metabolic Disease, 2012, 35, 51-63.	3.6	41
133	Drug dosing error with dropsâ€"severe clinical course of codeine intoxication in twins. European Journal of Pediatrics, 2009, 168, 819-824.	2.7	39
134	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	3.2	39
135	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
136	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
137	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
138	Mucolipidosis II presenting as severe neonatal hyperparathyroidism. European Journal of Pediatrics, 2005, 164, 236-243.	2.7	37
139	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
140	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
141	Bone and connective tissue disorders caused by defects in glycosaminoglycan biosynthesis: a panoramic view. FEBS Journal, 2019, 286, 3008-3032.	4.7	37
142	Spondyloperipheral dysplasia is caused by truncating mutations in the C-propeptide of COL2A1. , 2004, 129A, 144-148.		36
143	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
144	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

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145	StÃ¼veâ€“Wiedemann syndrome: longâ€term followâ€up and genetic heterogeneity. Clinical Genetics, 2010, 77, 266-272.	2.0	33
146	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
147	Analysis of the CTNS gene in patients of German and Swiss origin with nephropathic cystinosis. Human Mutation, 2002, 20, 237-237.	2.5	32
148	COL2A1â€“related skeletal dysplasias with predominant metaphyseal involvement. American Journal of Medical Genetics, Part A, 2007, 143A, 161-167.	1.2	32
149	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
150	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
151	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
152	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
153	Morphological, clinical and genetic aspects in a family with a novel LAMP-2 gene mutation (Danon) Tj ETQq1 1 0.784314 rgBT /Overlock	0.6	30
154	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
155	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
156	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
157	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. JCI Insight, 2019, 4, .	5.0	30
158	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
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