Stefan Mundlos

List of Publications by Year
in descending order

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305	30,872	4658 85	6130 159
papers	citations	h-index	g-index

327	327	327	35221
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cbfa1, a Candidate Gene for Cleidocranial Dysplasia Syndrome, Is Essential for Osteoblast Differentiation and Bone Development. Cell, 1997, 89, 765-771.	28.9	2,620
2	The single-cell transcriptional landscape of mammalian organogenesis. Nature, 2019, 566, 496-502.	27.8	2,292
3	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
4	The Human Phenotype Ontology: A Tool for Annotating and Analyzing Human Hereditary Disease. American Journal of Human Genetics, 2008, 83, 610-615.	6.2	797
5	The receptor tyrosine kinase Ror2 is involved in nonâ€canonical Wnt5a/JNK signalling pathway. Genes To Cells, 2003, 8, 645-654.	1.2	651
6	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	27.8	582
7	A High-Resolution Anatomical Atlas of the Transcriptome in the Mouse Embryo. PLoS Biology, 2011, 9, e1000582.	5.6	552
8	Structural variation in the 3D genome. Nature Reviews Genetics, 2018, 19, 453-467.	16.3	508
9	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	1.2	453
10	Clinical Diagnostics in Human Genetics with Semantic Similarity Searches in Ontologies. American Journal of Human Genetics, 2009, 85, 457-464.	6.2	444
11	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
12	Regulation of chondrocyte differentiation by Cbfa1. Mechanisms of Development, 1999, 80, 159-170.	1.7	424
13	Breaking TADs: How Alterations of Chromatin Domains Result in Disease. Trends in Genetics, 2016, 32, 225-237.	6.7	370
14	Classic and atypical fibrodysplasia ossificans progressiva (FOP) phenotypes are caused by mutations in the bone morphogenetic protein (BMP) type I receptor ACVR1. Human Mutation, 2009, 30, 379-390.	2.5	364
15	Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nature Genetics, 2008, 40, 32-34.	21.4	330
16	Mutations in the Transmembrane Natriuretic Peptide Receptor NPR-B Impair Skeletal Growth and Cause Acromesomelic Dysplasia, Type Maroteaux. American Journal of Human Genetics, 2004, 75, 27-34.	6.2	325
17	Plant nitric oxide synthase: a never-ending story?. Trends in Plant Science, 2006, 11, 524-525.	8.8	297
18	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopeniaâ€“Absent Radius Syndrome. American Journal of Human Genetics, 2007, 80, 232-240.	6.2	290

#	Article	IF	Citations
19	Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. Nature Genetics, 2010, 42, 827-829.	21.4	286
20	A restricted spectrum of NRAS mutations causes Noonan syndrome. Nature Genetics, 2010, 42, 27-29.	21.4	271
21	The Human Phenotype Ontology. Clinical Genetics, 2010, 77, 525-534.	2.0	267
22	Mutations in theRUNX2 gene in patients with cleidocranial dysplasia. Human Mutation, 2002, 19, 209-216.	2.5	263
23	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	6.2	240
24	Role of Runx Genes in Chondrocyte Differentiation. Developmental Biology, 2002, 245, 95-108.	2.0	233
25	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	12.4	223
26	Functional dissection of the Sox9â€“Kcnj2 locus identifies nonessential and instructive roles of TAD architecture. Nature Genetics, 2019, 51, 1263-1271.	21.4	223
27	Primary ciliary dyskinesia associated with normal axoneme ultrastructure is caused byDNAH11mutations. Human Mutation, 2008, 29, 289-298.	2.5	222
28	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
29	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
30	Mutation Analysis of Core Binding Factor A1 in Patients with Cleidocranial Dysplasia. American Journal of Human Genetics, 1999, 65, 1268-1278.	6.2	193
31	Activating and deactivating mutations in the receptor interaction site of GDF5 cause symphalangism or brachydactyly type A2. Journal of Clinical Investigation, 2005, 115, 2373-2381.	8.2	192
32	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. Journal of Experimental Medicine, 2013, 210, 433-443.	8.5	186
33	Deletions, Inversions, Duplications: Engineering of Structural Variants using CRISPR/Cas in Mice. Cell Reports, 2015, 10, 833-839.	6.4	181
34	Kniest and Stickler dysplasia phenotypes caused by collagen type II gene (COL2A1) defect. Nature Genetics, 1993, 3, 323-326.	21.4	179
35	Polymer physics predicts the effects of structural variants on chromatin architecture. Nature Genetics, 2018, 50, 662-667.	21.4	179
36	The fibrodysplasia ossificans progressiva R206H ACVR1 mutation activates BMP-independent chondrogenesis and zebrafish embryo ventralization. Journal of Clinical Investigation, 2009, 119, 3462-72.	8.2	178

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37	Comprehensive expression analysis of all Wnt genes and their major secreted antagonists during mouse limb development and cartilage differentiation. Gene Expression Patterns, 2009, 9, 215-223.	0.8	173
38	Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. Journal of Medical Genetics, 2006, 44, 131-135.	3.2	170
39	Loss of chondroitin 6-O-sulfotransferase-1 function results in severe human chondrodysplasia with progressive spinal involvement. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 10155-10160.	7.1	169
40	Distinct Mutations in the Receptor Tyrosine Kinase Gene ROR2 Cause Brachydactyly Type B. American Journal of Human Genetics, 2000, 67, 822-831.	6.2	166
41	Mutations in bone morphogenetic protein receptor 1B cause brachydactyly type A2. Proceedings of the National Academy of Sciences of the United States of America, 2003, 100, 12277-12282.	7.1	161
42	Genetic Disorders of the Skeleton: A Developmental Approach. American Journal of Human Genetics, 2003, 73, 447-474.	6.2	158
43	The other trinucleotide repeat: polyalanine expansion disorders. Current Opinion in Genetics and Development, 2005, 15, 285-293.	3.3	156
44	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	2.5	156
45	Mesenchymal stromal cells of myelodysplastic syndrome and acute myeloid leukemia patients have distinct genetic abnormalities compared with leukemic blasts. Blood, 2011, 118, 5583-5592.	1.4	150
46	Dynamic 3D chromatin architecture contributes to enhancer specificity and limb morphogenesis. Nature Genetics, 2018, 50, 1463-1473.	21.4	147
47	Regulatory Landscaping: How Enhancer-Promoter Communication Is Sculpted in 3D. Molecular Cell, 2019, 74, 1110-1122.	9.7	147
48	Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal Î³ Subunit. American Journal of Human Genetics, 2006, 79, 303-312.	6.2	146
49	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	21.4	146
50	Mutations in WNT7A Cause a Range of Limb Malformations, Including Fuhrmann Syndrome and Al-Awadi/Raas-Rothschild/Schinzel Phocomelia Syndrome. American Journal of Human Genetics, 2006, 79, 402-408.	6.2	144
51	Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423.	8.8	144
52	Identifying cis Elements for Spatiotemporal Control of Mammalian DNA Replication. Cell, 2019, 176, 816-830.e18.	28.9	144
53	A molecular pathogenesis for transcription factor associated poly-alanine tract expansions. Human Molecular Genetics, 2004, 13, 2351-2359.	2.9	139
54	Duplications Involving a Conserved Regulatory Element Downstream of BMP2 Are Associated with Brachydactyly Type A2. American Journal of Human Genetics, 2009, 84, 483-492.	6.2	139

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55	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	21.4	138
56	Mutations in PIGO, a Member of the GPI-Anchor-Synthesis Pathway, Cause Hyperphosphatasia with Mental Retardation. American Journal of Human Genetics, 2012, 91, 146-151.	6.2	135
57	TCR Repertoire Analysis by Next Generation Sequencing Allows Complex Differential Diagnosis of T Cellâ€"Related Pathology. American Journal of Transplantation, 2013, 13, 2842-2854.	4.7	131
58	Preformed chromatin topology assists transcriptional robustness of Shh during limb development. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 12390-12399.	7.1	131
59	A novel COL1A1 mutation in infantile cortical hyperostosis (Caffey disease) expands the spectrum of collagen-related disorders. Journal of Clinical Investigation, 2005, 115, 1250-1257.	8.2	129
60	Deletion and Point Mutations of PTHLH Cause Brachydactyly Type E. American Journal of Human Genetics, 2010, 86, 434-439.	6.2	127
61	Lumbar disc degeneration is linked to a carbohydrate sulfotransferase 3 variant. Journal of Clinical Investigation, 2013, 123, 4909-4917.	8.2	126
62	Looking beyond the genes: the role of non-coding variants in human disease. Human Molecular Genetics, 2016, 25, R157-R165.	2.9	125
63	Acetylcholine Receptor Pathway Mutations Explain Various Fetal Akinesia Deformation Sequence Disorders. American Journal of Human Genetics, 2008, 82, 464-476.	6.2	124
64	Characterization of hundreds of regulatory landscapes in developing limbs reveals two regimes of chromatin folding. Genome Research, 2017, 27, 223-233.	5.5	123
65	ecDNA hubs drive cooperative intermolecular oncogene expression. Nature, 2021, 600, 731-736.	27.8	123
66	Mutations in NSD1 are responsible for Sotos syndrome, but are not a frequent finding in other overgrowth phenotypes. European Journal of Human Genetics, 2003, 11, 858-865.	2.8	122
67	CA8 Mutations Cause a Novel Syndrome Characterized by Ataxia and Mild Mental Retardation with Predisposition to Quadrupedal Gait. PLoS Genetics, 2009, 5, e1000487.	3.5	120
68	A microduplication of the long range SHH limb regulator (ZRS) is associated with triphalangeal thumb-polysyndactyly syndrome. Journal of Medical Genetics, 2008, 45, 370-375.	3.2	118
69	Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Human Molecular Genetics, 2009, 18, 2149-2165.	2.9	115
70	Unblending of Transcriptional Condensates in Human Repeat Expansion Disease. Cell, 2020, 181, 1062-1079.e30.	28.9	115
71	Multiple roles for neurofibromin in skeletal development and growth. Human Molecular Genetics, 2007, 16, 874-886.	2.9	114
72	SOS1 is the second most common Noonan gene but plays no major role in cardio-facio-cutaneous syndrome. Journal of Medical Genetics, 2007, 44, 651-656.	3.2	114

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73	Ror2knockout mouse as a model for the developmental pathology of autosomal recessive Robinow syndrome. Developmental Dynamics, 2004, 229, 400-410.	1.8	113
74	Homeotic Arm-to-Leg Transformation Associated with Genomic Rearrangements at the PITX1 Locus. American Journal of Human Genetics, 2012, 91, 629-635.	6.2	111
75	Distinct CDH3 mutations cause ectodermal dysplasia, ectrodactyly, macular dystrophy (EEM) Tj ETQq1 1 0.784314 rgBT /Overlock 10	3.2	108
76	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
77	Composition and dosage of a multipartite enhancer cluster control developmental expression of Ihh (Indian hedgehog). Nature Genetics, 2017, 49, 1539-1545.	21.4	107
78	Serial genomic inversions induce tissue-specific architectural stripes, gene misexpression and congenital malformations. Nature Cell Biology, 2019, 21, 305-310.	10.3	107
79	A New Subtype of Brachydactyly Type B Caused by Point Mutations in the Bone Morphogenetic Protein Antagonist NOGGIN. American Journal of Human Genetics, 2007, 81, 388-396.	6.2	106
80	Duplications of noncoding elements 5â€² of SOX9 are associated with brachydactyly-anonychia. Nature Genetics, 2009, 41, 862-863.	21.4	105
81	Enhancer hijacking determines extrachromosomal circular MYCN amplicon architecture in neuroblastoma. Nature Communications, 2020, 11, 5823.	12.8	104
82	MicroRNAs Differentially Expressed in Postnatal Aortic Development Downregulate Elastin via 3â€² UTR and Coding-Sequence Binding Sites. PLoS ONE, 2011, 6, e16250.	2.5	100
83	Modulation of GDF5/BRI-b signalling through interaction with the tyrosine kinase receptor Ror2. Genes To Cells, 2004, 9, 1227-1238.	1.2	98
84	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. American Journal of Human Genetics, 2010, 87, 265-273.	6.2	98
85	PGAP2 Mutations, Affecting the GPI-Anchor-Synthesis Pathway, Cause Hyperphosphatasia with Mental Retardation Syndrome. American Journal of Human Genetics, 2013, 92, 584-589.	6.2	98
86	The three-dimensional genome: regulating gene expression during pluripotency and development. Development (Cambridge), 2017, 144, 3646-3658.	2.5	96
87	A mutation in Ihh that causes digit abnormalities alters its signalling capacity and range. Nature, 2009, 458, 1196-1200.	27.8	89
88	Copy-Number Variations Involving the IHH Locus Are Associated with Syndactyly and Craniosynostosis. American Journal of Human Genetics, 2011, 88, 70-75.	6.2	89
89	Induction of Macrophage Chemotaxis by Aortic Extracts of the mgR Marfan Mouse Model and a GxxPG-Containing Fibrillin-1 Fragment. Circulation, 2006, 114, 1855-1862.	1.6	88
90	Mutations in PGAP3 Impair GPI-Anchor Maturation, Causing a Subtype of Hyperphosphatasia with Mental Retardation. American Journal of Human Genetics, 2014, 94, 278-287.	6.2	88

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91	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. American Journal of Human Genetics, 2020, 106, 872-884.	6.2	85
92	A homozygous BMPR1B mutation causes a new subtype of acromesomelic chondrodysplasia with genital anomalies. Journal of Medical Genetics, 2005, 42, 314-317.	3.2	84
93	Mutations in GDF5 Reveal a Key Residue Mediating BMP Inhibition by NOGGIN. PLoS Genetics, 2009, 5, e1000747.	3.5	84
94	Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. European Journal of Human Genetics, 2012, 20, 754-761.	2.8	84
95	GDF5 Is a Second Locus for Multiple-Synostosis Syndrome. American Journal of Human Genetics, 2006, 78, 708-712.	6.2	83
96	Detection of novel skeletogenesis target genes by comprehensive analysis of a Runx2â^'/â^' mouse model. Gene Expression Patterns, 2007, 7, 102-112.	0.8	82
97	Redundant function of the heparan sulfate 6â€Oâ€endosulfatases Sulf1 and Sulf2 during skeletal development. Developmental Dynamics, 2008, 237, 339-353.	1.8	82
98	Mechanism for Release of Alkaline Phosphatase Caused by Glycosylphosphatidylinositol Deficiency in Patients with Hyperphosphatasia Mental Retardation Syndrome. Journal of Biological Chemistry, 2012, 287, 6318-6325.	3.4	82
99	Strategies for exome and genome sequence data analysis in diseaseâ€gene discovery projects. Clinical Genetics, 2011, 80, 127-132.	2.0	81
100	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
101	Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. Brain, 2014, 137, 683-692.	7.6	80
102	Homeobox genes d11â€“d13 and a13 control mouse autopod cortical bone and joint formation. Journal of Clinical Investigation, 2010, 120, 1994-2004.	8.2	76
103	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
104	The brachydactylies: a molecular disease family. Clinical Genetics, 2009, 76, 123-136.	2.0	74
105	Expression of Galectin-3 in Skeletal Tissues Is Controlled by Runx2. Journal of Biological Chemistry, 2003, 278, 17360-17367.	3.4	73
106	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	3.8	73
107	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	21.4	73
108	Microduplications encompassing the Sonic hedgehog limb enhancer <scp>ZRS</scp> are associated with Haasâ€type polysyndactyly and Laurinâ€Sandrow syndrome. Clinical Genetics, 2014, 86, 318-325.	2.0	72

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109	Title is missing!. Nature Genetics, 2001, 28, 37-41.	21.4	71
110	RGD-containing fibrillin-1 fragments upregulate matrix metalloproteinase expression in cell culture: A potential factor in the pathogenesis of the Marfan syndrome. Human Genetics, 2005, 116, 51-61.	3.8	71
111	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
112	Regulation of cell polarity in the cartilage growth plate and perichondrium of metacarpal elements by HOXD13 and WNT5A. Developmental Biology, 2014, 385, 83-93.	2.0	69
113	Negative regulation of Wnt signaling mediated by CK1â€phosphorylated DishevelledviaRor2. FASEB Journal, 2010, 24, 2417-2426.	0.5	68
114	Association of the 867Asp variant of the human anion exchanger 3 gene with common subtypes of idiopathic generalized epilepsy. Epilepsy Research, 2002, 51, 249-255.	1.6	67
115	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67
116	An inversion involving the mouse Shh locus results in brachydactyly through dysregulation of Shh expression. Journal of Clinical Investigation, 2005, 115, 900-909.	8.2	67
117	Wnt1 is an Lrp5-independent bone-anabolic Wnt ligand. Science Translational Medicine, 2018, 10, .	12.4	66
118	MiR-497âˆ¼195 Cluster MicroRNAs Regulate Osteoblast Differentiation by Targeting BMP Signaling. Journal of Bone and Mineral Research, 2015, 30, 796-808.	2.8	65
119	The role of 3D chromatin domains in gene regulation: a multi-facetted view on genome organization. Current Opinion in Genetics and Development, 2020, 61, 1-8.	3.3	64
120	A novel R486Q mutation in BMPR1B resulting in either a brachydactyly type C/symphalangism-like phenotype or brachydactyly type A2. European Journal of Human Genetics, 2006, 14, 1248-1254.	2.8	63
121	Deletions in PITX1 cause a spectrum of lower-limb malformations including mirror-image polydactyly. European Journal of Human Genetics, 2012, 20, 705-708.	2.8	63
122	Brachydactyly type A2 associated with a defect in proGDF5 processing. Human Molecular Genetics, 2008, 17, 1222-1233.	2.9	61
123	Deletions of the RUNX2 gene are present in about 10% of individuals with cleidocranial dysplasia. Human Mutation, 2010, 31, E1587-E1593.	2.5	61
124	Structural variations, the regulatory landscape of the genome and their alteration in human disease. BioEssays, 2013, 35, 533-543.	2.5	61
125	Expression of Runx2 transcription factor in nonâ€skeletal tissues, sperm and brain. Journal of Cellular Physiology, 2008, 217, 511-517.	4.1	60
126	Evolution of a Core Gene Network for Skeletogenesis in Chordates. PLoS Genetics, 2008, 4, e1000025.	3.5	59

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127	Brachydactyly type C caused by a homozygous missense mutation in the prodomain of CDMP1. , 2004, 124A, 356-363.		58
128	Expression of Type XXIII Collagen mRNA and Protein. Journal of Biological Chemistry, 2006, 281, 21546-21557.	3.4	58
129	Neurofibromin (Nf1) is required for skeletal muscle development. Human Molecular Genetics, 2011, 20, 2697-2709.	2.9	58
130	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
131	Receptor tyrosine kinase-like orphan receptor 2 (ROR2) and Indian hedgehog regulate digit outgrowth mediated by the phalanx-forming region. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 14211-14216.	7.1	57
132	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
133	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
134	Multiexon Deletions in the Type I Collagen COL1A2 Gene in Osteogenesis Imperfecta Type. Journal of Biological Chemistry, 1996, 271, 21068-21074.	3.4	55
135	Omaniâ€type spondyloepiphyseal dysplasia with cardiac involvement caused by a missense mutation in CHST3. Clinical Genetics, 2009, 75, 375-383.	2.0	54
136	Copy-Number Variations, Noncoding Sequences, and Human Phenotypes. Annual Review of Genomics and Human Genetics, 2011, 12, 53-72.	6.2	53
137	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. Neurology, 2021, 96, e1770-e1782.	1.1	53
138	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
139	A fibrillin-1-fragment containing theÂelastin-binding-protein GxxPG consensus sequence upregulates matrix metalloproteinase-1: biochemical andÂcomputational analysis. Journal of Molecular and Cellular Cardiology, 2006, 40, 234-246.	1.9	52
140	Exome sequencing and CRISPR/Cas genome editing identify mutations of ZAK as a cause of limb defects in humans and mice. Genome Research, 2016, 26, 183-191.	5.5	52
141	Mutant Hoxd13 induces extra digits in a mouse model of synpolydactyly directly and by decreasing retinoic acid synthesis. Journal of Clinical Investigation, 2009, 119, 146-56.	8.2	52
142	Mechanisms of digit formation: Human malformation syndromes tell the story. Developmental Dynamics, 2011, 240, 990-1004.	1.8	51
143	Whole exome sequencing identified a novel zinc-finger geneZNF141associated with autosomal recessive postaxial polydactyly type A. Journal of Medical Genetics, 2013, 50, 47-53.	3.2	51
144	Three-dimensional chromatin in disease: What holds us together and what drives us apart?. Current Opinion in Cell Biology, 2020, 64, 1-9.	5.4	51

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145	A Recurrent RNA-Splicing Mutation in the SEDL Gene Causes X-Linked Spondyloepiphyseal Dysplasia Tarda. American Journal of Human Genetics, 2001, 68, 1398-1407.	6.2	49
146	Cerebellar hypoplasia, with quadrupedal locomotion, caused by mutations in the very low-density lipoprotein receptor gene. European Journal of Human Genetics, 2008, 16, 1070-1074.	2.8	49
147	Wnt-ligand-dependent interaction of TAK1 (TGF-Î²-activated kinase-1) with the receptor tyrosine kinase Ror2 modulates canonical Wnt-signalling. Cellular Signalling, 2008, 20, 2134-2144.	3.6	48
148	Polyalanine expansion in HOXA13: three new affected families and the molecular consequences in a mouse model. Human Molecular Genetics, 2004, 13, 2841-2851.	2.9	47
149	A further case of the recurrent 15q24 microdeletion syndrome, detected by array CGH. European Journal of Pediatrics, 2008, 167, 903-908.	2.7	47
150	Homozygous deletion of chromosome 15q13.3 including CHRNA7 causes severe mental retardation, seizures, muscular hypotonia, and the loss of KLF13 and TRPM1 potentially cause macrocytosis and congenital retinal dysfunction in siblings. European Journal of Medical Genetics, 2011, 54, e441-e445.	1.3	47
151	Severe cleidocranial dysplasia can mimic hypophosphatasia. European Journal of Pediatrics, 2002, 161, 623-626.	2.7	46
152	Phenotypic variant of Brachydactyly-mental retardation syndrome in a family with an inherited interstitial 2q37.3 microdeletion including HDAC4. European Journal of Human Genetics, 2013, 21, 743-748.	2.8	46
153	Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Human Mutation, 2016, 37, 737-744.	2.5	46
154	Breakpoints around the HOXD cluster result in various limb malformations. Journal of Medical Genetics, 2005, 43, 111-118.	3.2	44
155	The synpolydactyly homolog (spdh) mutation in the mouse â€“ a defect in patterning and growth of limb cartilage elements. Mechanisms of Development, 2002, 112, 53-67.	1.7	43
156	Deletions of exons with regulatory activity at the DYNC1I1 locus are associated with split-hand/split-foot malformation: array CGH screening of 134 unrelated families. Orphanet Journal of Rare Diseases, 2014, 9, 108.	2.7	43
157	Missense variant in CCDC22 causes X-linked recessive intellectual disability with features of Ritscher-Schinzel/3C syndrome. European Journal of Human Genetics, 2015, 23, 633-638.	2.8	42
158	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
159	Spondyloepiphyseal dysplasia omani type: A new recessive type of SED with progressive spinal involvement. American Journal of Medical Genetics Part A, 2004, 126A, 413-419.	2.4	41
160	Ulnarâ€“mammary syndrome with dysmorphic facies and mental retardation caused by a novel 1.28 Mb deletion encompassing the TBX3 gene. European Journal of Human Genetics, 2006, 14, 1274-1279.	2.8	41
161	Filtering for Compound Heterozygous Sequence Variants in Non-Consanguineous Pedigrees. PLoS ONE, 2013, 8, e70151.	2.5	41
162	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. Molecular Genetics and Metabolism, 2014, 112, 310-316.	1.1	41

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163	The mole genome reveals regulatory rearrangements associated with adaptive intersexuality. Science, 2020, 370, 208-214.	12.6	41
164	The allele distribution in next-generation sequencing data sets is accurately described as the result of a stochastic branching process. Nucleic Acids Research, 2012, 40, 2426-2431.	14.5	40
165	Advances in computerâ€assisted syndrome recognition by the example of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 533-539.	3.6	40
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