Stefan Mundlos

List of Publications by Year
in descending order

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305 papers	30,872 citations	4658 85 h-index	6131 159 g-index
327 all docs	327 docs citations	327 times ranked	35221 citing authors

#	Article	IF	Citations
1	Cell adhesion and immune response, two main functions altered in the transcriptome of seasonally regressed testes of two mammalian species. Journal of Experimental Zoology Part B: Molecular and Developmental Evolution, 2023, 340, 231-244.	1.3	1
2	Biallelic truncating variants in ATP9A cause a novel neurodevelopmental disorder involving postnatal microcephaly and failure to thrive. Journal of Medical Genetics, 2022, 59, 662-668.	3.2	9
3	3D or Not 3D: Shaping the Genome during Development. Cold Spring Harbor Perspectives in Biology, 2022, 14, a040188.	5.5	11
4	Combining callers improves the detection of copy number variants from whole-genome sequencing. European Journal of Human Genetics, 2022, 30, 178-186.	2.8	18
5	Distal and proximal cis-regulatory elements sense X chromosome dosage and developmental state at the Xist locus. Molecular Cell, 2022, 82, 190-208.e17.	9.7	23
6	Xq27.1 palindrome mediated interchromosomal insertion likely causes familial congenital bilateral laryngeal abductor paralysis (Plott syndrome). Journal of Human Genetics, 2022, 67, 405-410.	2.3	3
7	<scp>LRFN5</scp> locus structure is associated with autism and influenced by the sex of the individual and locus conversions. Autism Research, 2022, 15, 421-433.	3.8	9
8	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	21.4	73
9	TADAâ€”a machine learning tool for functional annotation-based prioritisation of pathogenic CNVs. Genome Biology, 2022, 23, 67.	8.8	4
10	Genetic Diagnostics in Routine Osteological Assessment of Adult Low Bone Mass Disorders. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e3048-e3057.	3.6	12
11	Polycomb-mediated genome architecture enables long-range spreading of H3K27 methylation. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	33
12	EPEN-18. Oncogenic 3D genome conformations identify novel therapeutic targets in ependymoma. Neuro-Oncology, 2022, 24, i42-i42.	1.2	0
13	Expanding the clinical and molecular spectrum of <scp>ATP6V1A</scp> related metabolic cutis laxa. Journal of Inherited Metabolic Disease, 2021, 44, 972-986.	3.6	7
14	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
15	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. Nature, 2021, 592, 93-98.	27.8	53
16	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
17	Genome sequencing in families with congenital limb malformations. Human Genetics, 2021, 140, 1229-1239.	3.8	13
18	Relevant genetic variants are common in women with pregnancy and lactation-associated osteoporosis (PLO) and predispose to more severe clinical manifestations. Bone, 2021, 147, 115911.	2.9	14

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19	Complete lung agenesis caused by complex genomic rearrangements with neo-TAD formation at the SHH locus. Human Genetics, 2021, 140, 1459-1469.	3.8	9
20	One Gene, Many Facets: Multiple Immune Pathway Dysregulation in SOCS1 Haploinsufficiency. Frontiers in Immunology, 2021, 12, 680334.	4.8	11
21	GLI3 variants causing isolated polysyndactyly are not restricted to the protein's Câ€terminal third. Clinical Genetics, 2021, 100, 758-765.	2.0	4
22	Position effects at the FGF8 locus are associated with femoral hypoplasia. American Journal of Human Genetics, 2021, 108, 1725-1734.	6.2	4
23	A CRISPR-Cas9â€“engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	8
24	ecDNA hubs drive cooperative intermolecular oncogene expression. Nature, 2021, 600, 731-736.	27.8	123
25	What can go wrong in the non-coding genome and how to interpret whole genome sequencing data. Medizinische Genetik, 2021, 33, 121-131.	0.2	4
26	Split hand/foot malformation associated with 20p12.1 deletion: A case report. European Journal of Medical Genetics, 2020, 63, 103805.	1.3	1
27	The mole genome reveals regulatory rearrangements associated with adaptive intersexuality. Science, 2020, 370, 208-214.	12.6	41
28	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
29	Enhancer hijacking determines extrachromosomal circular MYCN amplicon architecture in neuroblastoma. Nature Communications, 2020, 11, 5823.	12.8	104
30	Unblending of Transcriptional Condensates in Human Repeat Expansion Disease. Cell, 2020, 181, 1062-1079.e30.	28.9	115
31	Hi-C Identifies Complex Genomic Rearrangements and TAD-Shuffling in Developmental Diseases. American Journal of Human Genetics, 2020, 106, 872-884.	6.2	85
32	Variable pulmonary manifestations in Chitayat syndrome: Six additional affected individuals. American Journal of Medical Genetics, Part A, 2020, 182, 2068-2076.	1.2	4
33	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
34	Skeletal deterioration in COL2A1-related spondyloepiphyseal dysplasia occurs prior to osteoarthritis. Osteoarthritis and Cartilage, 2020, 28, 334-343.	1.3	14
35	VarFish: comprehensive DNA variant analysis for diagnostics and research. Nucleic Acids Research, 2020, 48, W162-W169.	14.5	39
36	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

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37	Clinical Phenotype and Relevance of LRP5 and LRP6 Variants in Patients With Early-Onset Osteoporosis (EOOP). Journal of Bone and Mineral Research, 2020, 36, 271-282.	2.8	32
38	Efficiency of Computer-Aided Facial Phenotyping (DeepGestalt) in Individuals With and Without a Genetic Syndrome: Diagnostic Accuracy Study. Journal of Medical Internet Research, 2020, 22, e19263.	4.3	26
39	EPEN-04. ONCOGENIC 3D TUMOR GENOME ORGANIZATION IDENTIFIES NEW THERAPEUTIC TARGETS IN EPENDYMOMA. Neuro-Oncology, 2020, 22, iii308-iii308.	1.2	0
40	Integration of Hi-C and Nanopore Sequencing for Structural Variant Analysis in AML with a Complex Karyotype: (Chromothripsis)Â². Blood, 2020, 136, 28-28.	1.4	3
41	Jumping retroviruses nudge TADs apart. Nature Genetics, 2019, 51, 1304-1305.	21.4	3
42	Differentiation of MISSLA and Fanconi anaemia by computer-aided image analysis and presentation of two novel MISSLA siblings. European Journal of Human Genetics, 2019, 27, 1827-1835.	2.8	9
43	Functional dissection of the Sox9â€“Kcnj2 locus identifies nonessential and instructive roles of TAD architecture. Nature Genetics, 2019, 51, 1263-1271.	21.4	223
44	Nosology and classification of genetic skeletal disorders: 2019 revision. American Journal of Medical Genetics, Part A, 2019, 179, 2393-2419.	1.2	431
45	Lgr5 and Col22a1 Mark Progenitor Cells in the Lineage toward Juvenile Articular Chondrocytes. Stem Cell Reports, 2019, 13, 713-729.	4.8	35
46	H2AFY promoter deletion causes PITX1 endoactivation and Liebenberg syndrome. Journal of Medical Genetics, 2019, 56, 246-251.	3.2	20
47	Regulatory Landscaping: How Enhancer-Promoter Communication Is Sculpted in 3D. Molecular Cell, 2019, 74, 1110-1122.	9.7	147
48	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
49	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
50	GENE-02. CHROMOSOME CONFORMATION ANALYSIS OF EPENDYMOMA IDENTIFIES PUTATIVE TUMOR DEPENDENCY GENES ACTIVATED BY DISTAL ONCOGENIC ENHANCERS. Neuro-Oncology, 2019, 21, ii80-ii81.	1.2	0
51	SOPH syndrome in three affected individuals showing similarities with progeroid cutis laxa conditions in early infancy. Journal of Human Genetics, 2019, 64, 609-616.	2.3	14
52	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
53	Serial genomic inversions induce tissue-specific architectural stripes, gene misexpression and congenital malformations. Nature Cell Biology, 2019, 21, 305-310.	10.3	107
54	The single-cell transcriptional landscape of mammalian organogenesis. Nature, 2019, 566, 496-502.	27.8	2,292

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55	Identifying cis Elements for Spatiotemporal Control of Mammalian DNA Replication. Cell, 2019, 176, 816-830.e18.	28.9	144
56	Normal trabecular vertebral bone is formed via rapid transformation of mineralized spicules: A high-resolution 3D ex-vivo murine study. Acta Biomaterialia, 2019, 86, 429-440.	8.3	5
57	GOPHER: Generator Of Probes for capture Hi-C Experiments at high Resolution. BMC Genomics, 2019, 20, 40.	2.8	10
58	Structural variation in the 3D genome. Nature Reviews Genetics, 2018, 19, 453-467.	16.3	508
59	Polymer physics predicts the effects of structural variants on chromatin architecture. Nature Genetics, 2018, 50, 662-667.	21.4	179
60	Mutational analysis uncovers monogenic bone disorders in women with pregnancy-associated osteoporosis: three novel mutations in LRP5, COL1A1, and COL1A2. Osteoporosis International, 2018, 29, 1643-1651.	3.1	38
61	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
62	Transcriptional profiling of murine osteoblast differentiation based on RNA-seq expression analyses. Bone, 2018, 113, 29-40.	2.9	13
63	Response to Peron et al.. Genetics in Medicine, 2018, 20, 1481-1482.	2.4	2
64	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
65	A novel COL1A2 C-propeptide cleavage site mutation causing high bone mass osteogenesis imperfecta with a regional distribution pattern. Osteoporosis International, 2018, 29, 243-246.	3.1	12
66	Wnt1 is an Lrp5-independent bone-anabolic Wnt ligand. Science Translational Medicine, 2018, 10, .	12.4	66
67	Mutation in LBX1/Lbx1 precludes transcription factor cooperativity and causes congenital hypoventilation in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 13021-13026.	7.1	27
68	Dynamic 3D chromatin architecture contributes to enhancer specificity and limb morphogenesis. Nature Genetics, 2018, 50, 1463-1473.	21.4	147
69	Multisite de novo mutations in human offspring after paternal exposure to ionizing radiation. Scientific Reports, 2018, 8, 14611.	3.3	22
70	A novel mutation in CDH11, encoding cadherinâ€11, cause Branchioskeletogenital (Elsahyâ€Waters) syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2028-2033.	1.2	13
71	Comparison of Bone Microarchitecture Between Adult Osteogenesis Imperfecta and Early-Onset Osteoporosis. Calcified Tissue International, 2018, 103, 512-521.	3.1	29
72	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67

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73	Loss of murine Gfi1 causes neutropenia and induces osteoporosis depending on the pathogen load and systemic inflammation. PLoS ONE, 2018, 13, e0198510.	2.5	15
74	Impaired proteoglycan glycosylation, elevated TGF-Î² signaling, and abnormal osteoblast differentiation as the basis for bone fragility in a mouse model for gerodermia osteodysplastica. PLoS Genetics, 2018, 14, e1007242.	3.5	36
75	Novel splice mutation in LRP4 causes severe type of Cenani-Lenz syndactyly syndrome with oro-facial and skeletal symptoms. European Journal of Medical Genetics, 2017, 60, 421-425.	1.3	13
76	Characterization of hundreds of regulatory landscapes in developing limbs reveals two regimes of chromatin folding. Genome Research, 2017, 27, 223-233.	5.5	123
77	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
78	The three-dimensional genome: regulating gene expression during pluripotency and development. Development (Cambridge), 2017, 144, 3646-3658.	2.5	96
79	Mutations inMYO1Hcause a recessive form of central hypoventilation with autonomic dysfunction. Journal of Medical Genetics, 2017, 54, 754-761.	3.2	21
80	A Novel de novo FZD2 Mutation in a Patient with Autosomal Dominant Omodysplasia. Molecular Syndromology, 2017, 8, 318-324.	0.8	16
81	A likelihood ratio-based method to predict exact pedigrees for complex families from next-generation sequencing data. Bioinformatics, 2017, 33, 72-78.	4.1	8
82	A de novo 1q23.3-q24.2 deletion combined with a GORAB missense mutation causes a distinctive phenotype with cutis laxa. Journal of Human Genetics, 2017, 62, 325-328.	2.3	4
83	Composition and dosage of a multipartite enhancer cluster control developmental expression of Ihh (Indian hedgehog). Nature Genetics, 2017, 49, 1539-1545.	21.4	107
84	Genome-Wide Binding of Posterior HOXA/D Transcription Factors Reveals Subgrouping and Association with CTCF. PLoS Genetics, 2017, 13, e1006567.	3.5	38
85	Identification of a molecular defect in a stillborn fetus with perinatal lethal hypophosphatasia using a disease-associated genome sequencing approach. Polish Journal of Pathology, 2016, 1, 78-83.	0.3	3
86	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
87	Femoral facial syndrome associated with a de novo complex chromosome 2q37 rearrangement. American Journal of Medical Genetics, Part A, 2016, 170, 1202-1207.	1.2	9
88	Formation of new chromatin domains determines pathogenicity of genomic duplications. Nature, 2016, 538, 265-269.	27.8	582
89	The progressive ankylosis protein ANK facilitates clathrin- and adaptor-mediated membrane traffic at the trans-Golgi network-to-endosome interface. Human Molecular Genetics, 2016, 25, 3836-3848.	2.9	10
90	A homozygous HOXD13 missense mutation causes a severe form of synpolydactyly with metacarpal to carpal transformation. American Journal of Medical Genetics, Part A, 2016, 170, 615-621.	1.2	14

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91	Looking beyond the genes: the role of non-coding variants in human disease. Human Molecular Genetics, 2016, 25, R157-R165.	2.9	125
92	An overlapping phenotype of Osteogenesis imperfecta and Ehlersâ€“Danlos syndrome due to a heterozygous mutation in COL1A1 and biallelic missense variants in TNXB identified by whole exome sequencing. American Journal of Medical Genetics, Part A, 2016, 170, 1080-1085.	1.2	15
93	Duplication of PTHLH causes osteochondroplasia with a combined brachydactyly type E/A1 phenotype with disturbed bone maturation and rhizomelia. European Journal of Human Genetics, 2016, 24, 1132-1136.	2.8	8
94	Breaking TADs: How Alterations of Chromatin Domains Result in Disease. Trends in Genetics, 2016, 32, 225-237.	6.7	370
95	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
96	Comparison of Exome and Genome Sequencing Technologies for the Complete Capture of Proteinâ€Coding Regions. Human Mutation, 2015, 36, 815-822.	2.5	156
97	Nosology and classification of genetic skeletal disorders: 2015 revision. American Journal of Medical Genetics, Part A, 2015, 167, 2869-2892.	1.2	453
98	GORAB Missense Mutations Disrupt RAB6 and ARF5 Binding and Golgi Targeting. Journal of Investigative Dermatology, 2015, 135, 2368-2376.	0.7	28
99	Improved bone defect healing by a superagonistic GDF5 variant derived from a patient with multiple synostoses syndrome. Bone, 2015, 73, 111-119.	2.9	12
100	Deletions, Inversions, Duplications: Engineering of Structural Variants using CRISPR/Cas in Mice. Cell Reports, 2015, 10, 833-839.	6.4	181
101	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
102	Microdeletions on 6p22.3 are associated with mesomelic dysplasia Savarirayan type. Journal of Medical Genetics, 2015, 52, 476-483.	3.2	27
103	High resolution 3D laboratory x-ray tomography data of femora from young, 1â€“14 day old C57BL/6 mice. Data in Brief, 2015, 4, 32-33.	1.0	3
104	FGFR2 mutation in a patient without typical features of Pfeiffer syndrome â€“ The emerging role of combined NGS and phenotype based strategies. European Journal of Medical Genetics, 2015, 58, 376-380.	1.3	9
105	DOCK6Mutations Are Responsible for a Distinct Autosomal-Recessive Variant of Adams-Oliver Syndrome Associated with Brain and Eye Anomalies. Human Mutation, 2015, 36, 593-598.	2.5	32
106	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. European Journal of Human Genetics, 2015, 23, 870-873.	2.8	20
107	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. Nature Genetics, 2015, 47, 647-653.	21.4	146
108	Clinical Exome/Genome Reports-Announcement. Clinical Genetics, 2015, 87, 99-99.	2.0	0

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109	Disruptions of Topological Chromatin Domains Cause Pathogenic Rewiring of Gene-Enhancer Interactions. Cell, 2015, 161, 1012-1025.	28.9	1,725
110	Long bone maturation is driven by pore closing: A quantitative tomography investigation of structural formation in young C57BL/6 mice. Acta Biomaterialia, 2015, 22, 92-102.	8.3	20
111	Brachydactyly Type C patient with compound heterozygosity for p.Gly319Val and p.Ile358Thr variants in the GDF5 proregion: benign variants or mutations?. Journal of Human Genetics, 2015, 60, 419-425.	2.3	4
112	Strategies to improve the performance of rare variant association studies by optimizing the selection of controls. Bioinformatics, 2015, 31, btv457.	4.1	0
113	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
114	MiR-497â^¼195 Cluster MicroRNAs Regulate Osteoblast Differentiation by Targeting BMP Signaling. Journal of Bone and Mineral Research, 2015, 30, 796-808.	2.8	65
115	Double NF1 Inactivation Affects Adrenocortical Function in NF1Prx1 Mice and a Human Patient. PLoS ONE, 2015, 10, e0119030.	2.5	10
116	Up-regulation ofRUNX2in acute myeloid leukemia in a patient with an inherentRUNX2haploinsufficiency and cleidocranial dysplasia. Leukemia and Lymphoma, 2014, 55, 1930-1932.	1.3	12
117	Homozygous missense and nonsense mutations in BMPR1B cause acromesomelic chondrodysplasia-type Grebe. European Journal of Human Genetics, 2014, 22, 726-733.	2.8	23
118	The Liebenberg syndrome: in depth analysis of the original family. Journal of Hand Surgery: European Volume, 2014, 39, 919-925.	1.0	7
119	Screening for single nucleotide variants, small indels and exon deletions with a nextâ€generation sequencing based gene panel approach for <scp>U</scp> sher syndrome. Molecular Genetics & Genomic Medicine, 2014, 2, 393-401.	1.2	22
120	Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. Brain, 2014, 137, 683-692.	7.6	80
121	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
122	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
123	Deletions of chromosomal regulatory boundaries are associated with congenital disease. Genome Biology, 2014, 15, 423.	8.8	144
124	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	12.4	223
125	A novel mutation (g.106737G>T) in zone of polarizing activity regulatory sequence (ZRS) causes variable limb phenotypes in Werner mesomelia. American Journal of Medical Genetics, Part A, 2014, 164, 898-906.	1.2	15
126	Molecular Analysis of Two Novel Missense Mutations in the GDF5 Proregion That Reduce Protein Activity and Are Associated with Brachydactyly Type C. Journal of Molecular Biology, 2014, 426, 3221-3231.	4.2	10

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127	Neurofibromin inactivation impairs osteocyte development in Nf1Prx1 and Nf1Col1 mouse models. Bone, 2014, 66, 155-162.	2.9	6
128	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
129	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
130	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
131	Severe congenital cutis laxa with cardiovascular manifestations due to homozygous deletions in ALDH18A1. Molecular Genetics and Metabolism, 2014, 112, 310-316.	1.1	41
132	Multiscale, Converging Defects of Macro-Porosity, Microstructure and Matrix Mineralization Impact Long Bone Fragility in NF1. PLoS ONE, 2014, 9, e86115.	2.5	29
133	Brachydactyly Type B1. , 2014, , 92-94.		0
134	Catelâ€“Manzke Syndrome. , 2014, , 108-109.		0
135	Genotypeâ€“phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Molecular Genetics and Metabolism, 2013, 110, 352-361.	1.1	57
136	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
137	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
138	Severe neuronopathic autosomal recessive osteopetrosis due to homozygous deletions affecting OSTM1. Bone, 2013, 55, 292-297.	2.9	22
139	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
140	Mutations in WNT1 Cause Different Forms of Bone Fragility. American Journal of Human Genetics, 2013, 92, 565-574.	6.2	240
141	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
142	Structural variations, the regulatory landscape of the genome and their alteration in human disease. BioEssays, 2013, 35, 533-543.	2.5	61
143	Genome-wide linkage analysis is a powerful prenatal diagnostic tool in families with unknown genetic defects. European Journal of Human Genetics, 2013, 21, 367-372.	2.8	7
144	A GDF5 Point Mutation Strikes Twice - Causing BDA1 and SYNS2. PLoS Genetics, 2013, 9, e1003846.	3.5	34

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145	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
146	Whole exome sequencing identifiesFGF16nonsense mutations as the cause of X-linked recessive metacarpal 4/5 fusion. Journal of Medical Genetics, 2013, 50, 579-584.	3.2	31
147	Whole-exome sequencing identifies a novel missense mutation in EDAR causing autosomal recessive hypohidrotic ectodermal dysplasia with bilateral amastia and palmoplantar hyperkeratosis. British Journal of Dermatology, 2013, 168, 1353-1356.	1.5	23
148	Distinct global shifts in genomic binding profiles of limb malformation-associated HOXD13 mutations. Genome Research, 2013, 23, 2091-2102.	5.5	31
149	Filtering for Compound Heterozygous Sequence Variants in Non-Consanguineous Pedigrees. PLoS ONE, 2013, 8, e70151.	2.5	41
150	Lumbar disc degeneration is linked to a carbohydrate sulfotransferase 3 variant. Journal of Clinical Investigation, 2013, 123, 4909-4917.	8.2	126
151	Proximal microdeletions and microduplications of 1q21.1 contribute to variable abnormal phenotypes. European Journal of Human Genetics, 2012, 20, 754-761.	2.8	84
152	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
153	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
154	Further characterization of ATP6V0A2-related autosomal recessive cutis laxa. Human Genetics, 2012, 131, 1761-1773.	3.8	73
155	Microduplications upstream ofMSX2are associated with a phenocopy of cleidocranial dysplasia. Journal of Medical Genetics, 2012, 49, 437-441.	3.2	12
156	Duplications ofBHLHA9are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. Journal of Medical Genetics, 2012, 49, 119-125.	3.2	81
157	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
158	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
159	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
160	Deterioration of fracture healing in the mouse model of NF1 long bone dysplasia. Bone, 2012, 51, 651-660.	2.9	23
161	Copy-Number Variations, Noncoding Sequences, and Human Phenotypes. Annual Review of Genomics and Human Genetics, 2011, 12, 53-72.	6.2	53
162	The face of Ulnar Mammary syndrome?. European Journal of Medical Genetics, 2011, 54, 301-305.	1.3	16

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