Cornelius F Boerkoel

List of Publications by Year
in descending order

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154 papers	8,611 citations	71102 41 h-index	51608 86 g-index
162 all docs	162 docs citations	162 times ranked	13096 citing authors

#	Article	IF	Citations
1	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
2	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
3	Mutation of TDP1, encoding a topoisomerase Iâ€“dependent DNA damage repair enzyme, in spinocerebellar ataxia with axonal neuropathy. Nature Genetics, 2002, 32, 267-272.	21.4	475
4	Mutations in ALMS1 cause obesity, type 2 diabetes and neurosensory degeneration in AlstrÃ¶m syndrome. Nature Genetics, 2002, 31, 74-78.	21.4	358
5	Mutant chromatin remodeling protein SMARCAL1 causes Schimke immuno-osseous dysplasia. Nature Genetics, 2002, 30, 215-220.	21.4	297
6	Mutations in the neurofilament light chain gene (NEFL) cause early onset severe Charcot-Marie-Tooth disease. Brain, 2003, 126, 590-597.	7.6	259
7	Charcotâ€Marieâ€Tooth disease and related neuropathies: Mutation distribution and genotypeâ€phenotype correlation. Annals of Neurology, 2002, 51, 190-201.	5.3	257
8	The National Institutes of Health Undiagnosed Diseases Program: insights into rare diseases. Genetics in Medicine, 2012, 14, 51-59.	2.4	254
9	GRIN2A mutation and earlyâ€onset epileptic encephalopathy: personalized therapy with memantine. Annals of Clinical and Translational Neurology, 2014, 1, 190-198.	3.7	248
10	NBEAL2 is mutated in gray platelet syndrome and is required for biogenesis of platelet Î±-granules. Nature Genetics, 2011, 43, 732-734.	21.4	223
11	Periaxin Mutations Cause Recessive Dejerine-Sottas Neuropathy. American Journal of Human Genetics, 2001, 68, 325-333.	6.2	205
12	Manifestations and treatment of Schimke immuno-osseous dysplasia: 14 new cases and a review of the literature. European Journal of Pediatrics, 2000, 159, 1-7.	2.7	147
13	Detecting false-positive signals in exome sequencing. Human Mutation, 2012, 33, 609-613.	2.5	137
14	Congenital hypomyelinating neuropathy, central dysmyelination, and Waardenburg-Hirschsprung disease: Phenotypes linked bySOX10 mutation. Annals of Neurology, 2002, 52, 836-842.	5.3	129
15	Spinocerebellar ataxia with axonal neuropathy: consequence of a Tdp1 recessive neomorphic mutation?. EMBO Journal, 2007, 26, 4732-4743.	7.8	129
16	Functional analysis of a de novo GRIN2A missense mutation associated with early-onset epileptic encephalopathy. Nature Communications, 2014, 5, 3251.	12.8	128
17	1,25-(OH)2D-24 Hydroxylase (CYP24A1) Deficiency as a Cause of Nephrolithiasis. Clinical Journal of the American Society of Nephrology: CJASN, 2013, 8, 649-657.	4.5	127
18	Rare Copy Number Variants Contribute to Congenital Left-Sided Heart Disease. PLoS Genetics, 2012, 8, e1002903.	3.5	119

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19	Glycosylation, Hypogammaglobulinemia, and Resistance to Viral Infections. New England Journal of Medicine, 2014, 370, 1615-1625.	27.0	117
20	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	2.5	107
21	Periaxin mutations cause a broad spectrum of demyelinating neuropathies. Annals of Neurology, 2002, 51, 709-715.	5.3	106
22	Genes in a Refined Smith-Magenis Syndrome Critical Deletion Interval on Chromosome 17p11.2 and the Syntenic Region of the Mouse. Genome Research, 2002, 12, 713-728.	5.5	101
23	York platelet syndrome is a CRAC channelopathy due to gain-of-function mutations in STIM1. Molecular Genetics and Metabolism, 2015, 114, 474-482.	1.1	94
24	Advances in chromatin remodeling and human disease. Current Opinion in Genetics and Development, 2004, 14, 308-315.	3.3	93
25	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
26	Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. Genetics in Medicine, 2016, 18, 608-617.	2.4	85
27	The Role of Nuclear Bodies in Gene Expression and Disease. Biology, 2013, 2, 976-1033.	2.8	71
28	Chromatin remodeling and human disease. Current Opinion in Genetics and Development, 2003, 13, 246-252.	3.3	70
29	Overexpression screens identify conserved dosage chromosome instability genes in yeast and human cancer. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 9967-9976.	7.1	67
30	CMT4A: Identification of a HispanicGDAP1 founder mutation. Annals of Neurology, 2003, 53, 400-405.	5.3	64
31	Hypomorphic Temperature-Sensitive Alleles of NSDHL Cause CK Syndrome. American Journal of Human Genetics, 2010, 87, 905-914.	6.2	64
32	Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome. Nature Communications, 2017, 8, 1257.	12.8	64
33	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
34	EGR2 mutation R359W causes a spectrum of Dejerine-Sottas neuropathy. Neurogenetics, 2001, 3, 153-157.	1.4	60
35	Three rare diseases in one Sib pair: RAI1, PCK1, GRIN2B mutations associated with Smithâ€“Magenis Syndrome, cytosolic PEPCK deficiency and NMDA receptor glutamate insensitivity. Molecular Genetics and Metabolism, 2014, 113, 161-170.	1.1	58
36	Penetrance of biallelic SMARCAL1 mutations is associated with environmental and genetic disturbances of gene expression. Human Molecular Genetics, 2012, 21, 2572-2587.	2.9	57

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37	The implications of familial incidental findings from exome sequencing: the NIH Undiagnosed Diseases Program experience. Genetics in Medicine, 2014, 16, 741-750.	2.4	56
38	Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. American Journal of Medical Genetics, Part A, 2016, 170, 2440-2444.	1.2	56
39	Exome sequencing and SNP analysis detect novel compound heterozygosity in fatty acid hydroxylase-associated neurodegeneration. European Journal of Human Genetics, 2012, 20, 476-479.	2.8	55
40	Schimke immunoosseous dysplasia: suggestions of genetic diversity. Human Mutation, 2007, 28, 273-283.	2.5	49
41	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Molecular Genetics and Metabolism, 2015, 115, 128-140.	1.1	44
42	Genetic defects in patients with glycogenosis type II (acid maltase deficiency). Muscle and Nerve, 1995, 18, S70-S74.	2.2	43
43	Schimke immuno-osseous dysplasia: SMARCAL1 loss-of-function and phenotypic correlation. Journal of Medical Genetics, 2008, 46, 49-59.	3.2	43
44	A Novel N-Tetrasaccharide in Patients with Congenital Disorders of Glycosylation, Including Asparagine-Linked Glycosylation Protein 1, Phosphomannomutase 2, and Mannose Phosphate Isomerase Deficiencies. Clinical Chemistry, 2016, 62, 208-217.	3.2	43
45	ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. PLoS Genetics, 2017, 13, e1006481.	3.5	42
46	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
47	Identification of a Putative Tdp1 Inhibitor (CD00509) by in Vitro and Cell-Based Assays. Journal of Biomolecular Screening, 2014, 19, 1372-1382.	2.6	41
48	A New Defective Retroviral Vector System Based on the Bryan Strain of Rous Sarcoma Virus. Virology, 1993, 195, 669-679.	2.4	39
49	AlstrÃ¶m syndrome: further evidence for linkage to human chromosome 2p13. Human Genetics, 1999, 105, 474-479.	3.8	39
50	SET binding factor 2 (SBF2) mutation causes CMT4B with juvenile onset glaucoma. Neurology, 2004, 63, 577-580.	1.1	39
51	Retroviral Transfer of AcidÎ±-Glucosidase cDNA to Enzyme-Deficient Myoblasts Results in Phenotypic Spread of the Genotypic Correction by Both Secretion and Fusion. Human Gene Therapy, 1997, 8, 1555-1563.	2.7	38
52	Disease tropism of c-erbB: effects of carboxyl-terminal tyrosine and internal mutations on tissue-specific transformation.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 7164-7168.	7.1	37
53	Longevity in Schimke immuno-osseous dysplasia. Journal of Medical Genetics, 2002, 39, 922-925.	3.2	37
54	Screening for mutations in a genetically heterogeneous disorder: DHPLC versus DNA sequence for mutation detection in multiple genes causing Charcot-Marie-Tooth neuropathy. Genetics in Medicine, 2001, 3, 335-342.	2.4	36

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55	Molecular Mechanisms for CMT1A Duplication and HNPP Deletion. Annals of the New York Academy of Sciences, 1999, 883, 22-35.	3.8	35
56	Frequent monoallelic loss of D13S319 in multiple myeloma patients shown by interphase fluorescence in situ hybridization. Leukemia, 1999, 13, 105-109.	7.2	35
57	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. Orphanet Journal of Rare Diseases, 2016, 11, 62.	2.7	35
58	SMARCAL1 deficiency predisposes to nonâ€Hodgkin lymphoma and hypersensitivity to genotoxic agents in vivo. American Journal of Medical Genetics, Part A, 2012, 158A, 2204-2213.	1.2	34
59	Analysis of DNA sequence variants detected by high-throughput sequencing. Human Mutation, 2012, 33, 599-608.	2.5	34
60	B-lymphoma induction by reticuloendotheliosis virus: characterization of a mutated chicken syncytial virus provirus involved in c-myc activation. Journal of Virology, 1987, 61, 2084-2090.	3.4	33
61	Transcriptional interaction between retroviral long terminal repeats (LTRs): mechanism of 5' LTR suppression and 3' LTR promoter activation of c-myc in avian B-cell lymphomas. Journal of Virology, 1992, 66, 4814-4823.	3.4	33
62	Denaturing high-performance liquid chromatography of the myotubularin-related 2 gene (MTMR2) in unrelated patients with Charcot-Marie-Tooth disease suggests a low frequency of mutation in inherited neuropathy. Neurogenetics, 2001, 3, 107-109.	1.4	32
63	Exome sequencing as a diagnostic tool in a case of undiagnosed juvenile-onset GM ₁ -gangliosidosis. Neurology, 2012, 79, 123-126.	1.1	32
64	SMARCAL1 and replication stress: An explanation for SIOD?. Nucleus, 2010, 1, 245-248.	2.2	31
65	Methylation profiling in individuals with Russellâ€“Silver syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 347-355.	1.2	31
66	TDP1 and PARP1 Deficiency Are Cytotoxic to Rhabdomyosarcoma Cells. Molecular Cancer Research, 2013, 11, 1179-1192.	3.4	31
67	Schimke immuno-osseous dysplasia: A cell autonomous disorder?. American Journal of Medical Genetics, Part A, 2006, 140A, 340-348.	1.2	30
68	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics, 2009, 10, 526.	2.8	30
69	Association of migraine-like headaches with Schimke immuno-osseous dysplasia. American Journal of Medical Genetics, Part A, 2005, 135A, 206-210.	1.2	29
70	An analysis of exome sequencing for diagnostic testing of the genes associated with muscle disease and spastic paraplegia. Human Mutation, 2012, 33, 614-626.	2.5	29
71	Schimke immunoosseous dysplasia complicated by moyamoya phenomenon. , 1998, 78, 118-122.		28
72	Schimke-immuno-osseous dysplasia: New mutation with weak genotype-phenotype correlation in siblings. American Journal of Medical Genetics, Part A, 2005, 135A, 202-205.	1.2	28

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73	Schimke Versus Non-Schimke Chronic Kidney Disease: An Anthropometric Approach. Pediatrics, 2006, 118, e400-e407.	2.1	28
74	The NIH Undiagnosed Diseases Program: bonding scientists and clinicians. DMM Disease Models and Mechanisms, 2012, 5, 3-5.	2.4	28
75	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	21.4	28
76	Panâ€cancer RNAâ€seq data stratifies tumours by some hallmarks of cancer. Journal of Cellular and Molecular Medicine, 2020, 24, 418-430.	3.6	28
77	Characterization of a new Xâ€linked mental retardation syndrome with microcephaly, cortical malformation, and thin habitus. American Journal of Medical Genetics, Part A, 2009, 149A, 2469-2478.	1.2	27
78	VAR-MD: A tool to analyze whole exome-genome variants in small human pedigrees with mendelian inheritance. Human Mutation, 2012, 33, 593-598.	2.5	27
79	Impaired osteoblast and osteoclast function characterize the osteoporosis of Snyder - Robinson syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 27.	2.7	27
80	Disruption of Golgi morphology and altered protein glycosylation in PLA2G6-associated neurodegeneration. Journal of Medical Genetics, 2016, 53, 180-189.	3.2	27
81	The genetic convergence of Charcot-Marie-Tooth disease types 1 and 2 and the role of genetics in sporadic neuropathy. Current Neurology and Neuroscience Reports, 2002, 2, 70-77.	4.2	26
82	Lethal neonatal Menkes' disease with severe vasculopathy and fractures. Acta Paediatrica, International Journal of Paediatrics, 1998, 87, 1297-1300.	1.5	26
83	Neurologic Phenotype of Schimke Immuno-Osseous Dysplasia and Neurodevelopmental Expression of SMARCAL1. Journal of Neuropathology and Experimental Neurology, 2008, 67, 565-577.	1.7	26
84	SMARCAL1 and replication stress. Nucleus, 2010, 1, 245-248.	2.2	26
85	Reduced elastogenesis: a clue to the arteriosclerosis and emphysematous changes in Schimke immuno-osseous dysplasia?. Orphanet Journal of Rare Diseases, 2012, 7, 70.	2.7	26
86	Gene Clusters, Molecular Evolution and Disease: A Speculation. Current Genomics, 2009, 10, 64-75.	1.6	25
87	Gray matter heterotopias and brachytelephalangic chondrodysplasia punctata: A complication of hyperemesis gravidarum induced vitamin K deficiency?. American Journal of Medical Genetics, Part A, 2007, 143A, 200-204.	1.2	24
88	Defining Disease, Diagnosis, and Translational Medicine within a Homeostatic Perturbation Paradigm: The National Institutes of Health Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2017, 4, 62.	2.6	23
89	Schimke immunoosseous dysplasia: defining skeletal features. European Journal of Pediatrics, 2010, 169, 801-811.	2.7	22
90	Rituximab resistant evans syndrome and autoimmunity in Schimke immuno-osseous dysplasia. Pediatric Rheumatology, 2011, 9, 27.	2.1	22

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91	Novel SNP array analysis and exome sequencing detect a homozygous exon 7 deletion of MEGF10 causing early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). Neuromuscular Disorders, 2013, 23, 483-488.	0.6	22
92	Lack of IL7RÎ± expression in T cells is a hallmark of T-cell immunodeficiency in Schimke immuno-osseous dysplasia (SIOD). Clinical Immunology, 2015, 161, 355-365.	3.2	22
93	MED23â€associated intellectual disability in a nonâ€consanguineous family. American Journal of Medical Genetics, Part A, 2015, 167, 1374-1380.	1.2	21
94	Expression profile and mitochondrial colocalization of Tdp1 in peripheral human tissues. Journal of Molecular Histology, 2013, 44, 481-494.	2.2	20
95	Recurrent subacute post-viral onset of ataxia associated with a PRF1 mutation. European Journal of Human Genetics, 2013, 21, 1232-1239.	2.8	19
96	Desmosterolosis: an illustration of diagnostic ambiguity of cholesterol synthesis disorders. Orphanet Journal of Rare Diseases, 2014, 9, 94.	2.7	19
97	Pharmacogenomic incidental findings in 308 families: The NIH Undiagnosed Diseases Program experience. Genetics in Medicine, 2016, 18, 1303-1307.	2.4	19
98	Clinical and genetic distinction of Schimke immunoâ€osseous dysplasia and cartilageâ€hair hypoplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 2013-2017.	1.2	18
99	Sensitive quantification of mosaicism using high density SNP arrays and the cumulative distribution function. Molecular Genetics and Metabolism, 2012, 105, 665-671.	1.1	18
100	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. American Journal of Medical Genetics, Part A, 2017, 173, 3172-3181.	1.2	18
101	Reactive oxygen species stress increases accumulation of tyrosyl-DNA phsosphodiesterase 1 within mitochondria. Scientific Reports, 2018, 8, 4304.	3.3	18
102	Recombination mapping using Boolean logic and high-density SNP genotyping for exome sequence filtering. Molecular Genetics and Metabolism, 2012, 105, 382-389.	1.1	17
103	Insights into the Renal Pathogenesis in Schimke Immuno-Osseous Dysplasia. Journal of Histochemistry and Cytochemistry, 2015, 63, 32-44.	2.5	17
104	Phenotypic evolution of UNC80 loss of function. American Journal of Medical Genetics, Part A, 2016, 170, 3106-3114.	1.2	17
105	Cerebellar atrophy in Schimkeâ€immunoâ€osseous dysplasia. American Journal of Medical Genetics, Part A, 2007, 143A, 2040-2045.	1.2	16
106	Bone marrow transplantation in Schimke immunoâ€osseous dysplasia. American Journal of Medical Genetics, Part A, 2013, 161, 2609-2613.	1.2	16
107	Increased Wnt and Notch signaling: a clue to the renal disease in Schimke immuno-osseous dysplasia?. Orphanet Journal of Rare Diseases, 2016, 11, 149.	2.7	16
108	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1450-1465.	6.2	16

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109	Human isoleucyl-tRNA synthetase: Sequence of the cDNA, alternative mRNA splicing, and the characteristics of an unusually long C-terminal extension. Gene, 1995, 155, 299-304.	2.2	15
110	ERCC6 dysfunction presenting as progressive neurological decline with brain hypomyelination. American Journal of Medical Genetics, Part A, 2014, 164, 2892-2900.	1.2	15
111	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. BMC Genomics, 2017, 18, 403.	2.8	15
112	Developmental expression pattern of the cholesterogenic enzyme NSDHL and negative selection of NSDHL-deficient cells in the heterozygous Bpa1H/+ mouse. Molecular Genetics and Metabolism, 2009, 98, 356-366.	1.1	14
113	19p13.2 microduplication causes a Sotos syndromeâ€like phenotype and alters gene expression. Clinical Genetics, 2012, 81, 56-63.	2.0	14
114	Mitotic Intragenic Recombination: A Mechanism of Survival for Several Congenital Disorders of Glycosylation. American Journal of Human Genetics, 2016, 98, 339-346.	6.2	14
115	Abnormal glycosylation in Joubert syndrome type 10. Cilia, 2017, 6, 2.	1.8	14
116	Improved outcome with immunosuppressive monotherapy after renal transplantation in Schimkeâ€immunoâ€osseous dysplasia. Pediatric Transplantation, 2009, 13, 482-489.	1.0	13
117	A NovelAMELXMutation, Its Phenotypic Features, and Skewed X Inactivation. Journal of Dental Research, 2019, 98, 870-878.	5.2	12
118	Fatal lymphoproliferative disorder in a child with Schimke immuno-osseous dysplasia. American Journal of Medical Genetics Part A, 2004, 131A, 194-199.	2.4	11
119	Vaso-occlusion in Schimke-immuno-osseous Dysplasia: Is the NO Pathway Involved?. Hormone and Metabolic Research, 2006, 38, 678-682.	1.5	10
120	A cryptic familial rearrangement of 11p15.5, involving both imprinting centers, in a family with a history of short stature. American Journal of Medical Genetics, Part A, 2014, 164, 1587-1594.	1.2	10
121	Spinocerebellar Ataxia with Axonal Neuropathy. Advances in Experimental Medicine and Biology, 2010, 685, 75-83.	1.6	10
122	Minimal genotypeâ€“phenotype correlation for small deletions within distal 1p36. American Journal of Medical Genetics, Part A, 2011, 155, 3164-3169.	1.2	9
123	Coffinâ€Siris syndrome: Phenotypic evolution of a novel SMARCA4 mutation. American Journal of Medical Genetics, Part A, 2014, 164, 1808-1814.	1.2	9
124	4q12â€“4q21.21 deletion genotypeâ€“phenotype correlation and the absence of piebaldism in presence of KIT haploinsufficiency. American Journal of Medical Genetics, Part A, 2015, 167, 231-237.	1.2	9
125	Transcriptional and posttranscriptional mechanisms contribute to the dysregulation of elastogenesis in Schimke immuno-osseous dysplasia. Pediatric Research, 2015, 78, 609-617.	2.3	9
126	Chromatin changes inSMARCAL1deficiency: A hypothesis for the gene expression alterations of Schimke immuno-osseous dysplasia. Nucleus, 2016, 7, 560-571.	2.2	9

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127	Kearns-Sayre syndrome presenting as isolated growth failure. BMJ Case Reports, 2013, 2013, bcr2012007272-bcr2012007272.	0.5	8
128	Glycomics in rare diseases: from diagnosis tomechanism. Translational Research, 2019, 206, 5-17.	5.0	8
129	Somatic mosaicism detected by genome-wide sequencing in 500 parentâ€“child trios with suspected genetic disease: clinical and genetic counseling implications. Journal of Physical Education and Sports Management, 2021, 7, a006125.	1.2	8
130	Expression profile of NSDHL in human peripheral tissues. Journal of Molecular Histology, 2012, 43, 95-106.	2.2	7
131	Congenital Bilateral Retinal Detachment in Two Siblings with Osteoporosis-Pseudoglioma Syndrome. Ophthalmic Genetics, 2015, 36, 276-280.	1.2	7
132	Genome-wide sequencing and the clinical diagnosis of genetic disease: The CAUSES study. Human Genetics and Genomics Advances, 2022, 3, 100108.	1.7	7
133	Beckwithâ€“Wiedemann syndrome in sibs discordant for IC2 methylation. American Journal of Medical Genetics, Part A, 2012, 158A, 1662-1669.	1.2	6
134	Replicate exome-sequencing in a multiple-generation family: improved interpretation of next-generation sequencing data. BMC Genomics, 2015, 16, 998.	2.8	6
135	Longâ€read genome sequencing resolves a complex 13q structural variant associated with syndromic anophthalmia. American Journal of Medical Genetics, Part A, 2022, 188, 1589-1594.	1.2	4
136	The Clinical Variant Analysis Tool: Analyzing the evidence supporting reported genomic variation in clinical practice. Genetics in Medicine, 2022, 24, 1512-1522.	2.4	4
137	Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. Frontiers in Medicine, 2016, 3, 39.	2.6	3
138	An approach to rapid characterization of DMD copy number variants for prenatal risk assessment. American Journal of Medical Genetics, Part A, 2021, 185, 2541-2545.	1.2	3
139	The National Institutes of Health Undiagnosed Diseases Program: Insights into rare diseases. Genetics in Medicine, 2011, , 1.	2.4	3
140	An infant with congenital respiratory insufficiency and diaphragmatic paralysis: A novel <scp>BICD2</scp> phenotype?. American Journal of Medical Genetics, Part A, 2022, 188, 926-930.	1.2	3
141	Can leaky splicing and evasion of premature termination codon surveillance contribute to the phenotypic variability in Alkuraya-Kucinskas syndrome?. European Journal of Medical Genetics, 2022, 65, 104427.	1.3	3
142	A novel 8.5 MB dup(1)(p34.1p34.3) characterized by FISH in a child presenting with congenital heart defect and dysmorphic features. American Journal of Medical Genetics, Part A, 2006, 140A, 1864-1870.	1.2	2
143	A novel syndrome with psychiatric features and review of malformation syndromes with psychiatric disorders. American Journal of Medical Genetics, Part A, 2009, 149A, 713-721.	1.2	2
144	Phenotypic and molecular characterization of a novel DCX deletion and a review of the literature. Clinical Genetics, 2009, 76, 214-218.	2.0	2

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145	Hypothesis: SLC12A3 Polymorphism modifies thiazide hypersensitivity of antenatal Bartter syndrome to thiazide resistance. European Journal of Medical Genetics, 2012, 55, 96-98.	1.3	2
146	Cultural differences define diagnosis and genomic medicine practice: implications for undiagnosed diseases program in China. Frontiers of Medicine, 2013, 7, 389-394.	3.4	2
147	Fetal alcohol syndrome: a phenocopy of spondylocarpotarsal synostosis syndrome?. Clinical Dysmorphology, 2010, 19, 175-180.	0.3	1
148	Importance of neurologic and cutaneous signs in the diagnosis of Schimke immuno-osseous dysplasia. Turkish Journal of Pediatrics, 2015, 57, 509-13.	0.6	1
149	IMPROVED OUTCOME WITH IMMUNOSUPPRESSIVE MONOTHERAPY AFTER RENAL TRANSPLANTATION IN SCHIMKE-IMMUNO-OSSEOUS DYSPLASIA. Transplantation, 2008, 86, 613.	1.0	0
150	Evaluating patients referred to a medical genetics connective tissue disorder clinic. Molecular Genetics and Metabolism, 2021, 132, S146.	1.1	0
151	Single center experience in the application of evidence based medicine to the clinical interpretation of genomic sequence reports. Molecular Genetics and Metabolism, 2021, 132, S167-S168.	1.1	0
152	Mesenteric cysts, lymphatic leak, and cerebral cavernous malformation in a proband with KRIT1 â€related disease. American Journal of Medical Genetics, Part A, 2021, , .	1.2	0
153	Atrophy of vermis cerebelli in Schimke-immuno-osseous dysplasia. Neuropediatrics, 2006, 37, .	0.6	0
154	Two Sisters with Congenital Blindness caused by Osteoporosis-pseudoglioma Syndrome due to new Mutations in the LPR5 Gene. Acta Ophthalmologica, 2015, 93, n/a-n/a.	1.1	0