James R Lupski

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2350399/publications.pdf

Version: 2024-02-01

457 papers	46,758 citations	2309 101 h-index	2970 195 g-index
473 all docs	473 docs citations	473 times ranked	45378 citing authors

#	Article	IF	Citations
1	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	13.9	1,717
2	The complete genome of an individual by massively parallel DNA sequencing. Nature, 2008, 452, 872-876.	13.7	1,635
3	DNA duplication associated with Charcot-Marie-Tooth disease type 1A. Cell, 1991, 66, 219-232.	13.5	1,313
4	A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Starqardt macular dystrophy. Nature Genetics, 1997, 15, 236-246.	9.4	1,277
5	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
6	Copy Number Variation in Human Health, Disease, and Evolution. Annual Review of Genomics and Human Genetics, 2009, 10, 451-481.	2.5	1,026
7	Structural Variation in the Human Genome and its Role in Disease. Annual Review of Medicine, 2010, 61, 437-455.	5.0	1,015
8	Mutation of the Stargardt Disease Gene (ABCR) in Age-Related Macular Degeneration. Science, 1997, 277, 1805-1807.	6.0	844
9	Genomic disorders: structural features of the genome can lead to DNA rearrangements and human disease traits. Trends in Genetics, 1998, 14, 417-422.	2.9	817
10	A DNA Replication Mechanism for Generating Nonrecurrent Rearrangements Associated with Genomic Disorders. Cell, 2007, 131, 1235-1247.	13.5	756
11	A Microhomology-Mediated Break-Induced Replication Model for the Origin of Human Copy Number Variation. PLoS Genetics, 2009, 5, e1000327.	1.5	700
12	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	13.9	698
13	Triallelic Inheritance in Bardet-Biedl Syndrome, a Mendelian Recessive Disorder. Science, 2001, 293, 2256-2259.	6.0	599
14	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
15	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
16	The gene for the peripheral myelin protein PMPâ€“22 is a candidate for Charcotâ€“Marieâ€“Tooth disease type 1A. Nature Genetics, 1992, 1, 159-165.	9.4	529
17	Mechanisms underlying structural variant formation in genomic disorders. Nature Reviews Genetics, 2016, 17, 224-238.	7.7	526
18	Genomic Disorders: Molecular Mechanisms for Rearrangements and Conveyed Phenotypes. PLoS Genetics, 2005, 1, e49.	1.5	496

#	Article	IF	Citations
19	Mutations in the early growth response 2 (EGR2) gene are associated with hereditary myelinopathies. Nature Genetics, 1998, 18, 382-384.	9.4	475
20	Non-coding genetic variants in human disease: Figure 1.. Human Molecular Genetics, 2015, 24, R102-R110.	1.4	466
21	Human Genome Sequencing in Health and Disease. Annual Review of Medicine, 2012, 63, 35-61.	5.0	404
22	Chromosome Catastrophes Involve Replication Mechanisms Generating Complex Genomic Rearrangements. Cell, 2011, 146, 889-903.	13.5	391
23	Charcotâ€“Marieâ€“Tooth type 1A duplication appears to arise from recombination at repeat sequences flanking the 1.5 Mb monomer unit. Nature Genetics, 1992, 2, 292-300.	9.4	385
24	Molecular mechanism for distinct neurological phenotypes conveyed by allelic truncating mutations. Nature Genetics, 2004, 36, 361-369.	9.4	383
25	The DNA replication FoSTeS/MMBIR mechanism can generate genomic, genic and exonic complex rearrangements in humans. Nature Genetics, 2009, 41, 849-853.	9.4	382
26	Charcot-Marie-Tooth Disease Type 1A -- Association with a Spontaneous Point Mutation in the PMP22 Gene. New England Journal of Medicine, 1993, 329, 96-101.	13.9	375
27	Genomic rearrangements and sporadic disease. Nature Genetics, 2007, 39, S43-S47.	9.4	373
28	Homologous recombination of a flanking repeat gene cluster is a mechanism for a common contiguous gene deletion syndrome. Nature Genetics, 1997, 17, 154-163.	9.4	364
29	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
30	Clan Genomics and the Complex Architecture of Human Disease. Cell, 2011, 147, 32-43.	13.5	330
31	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
32	Mutations in MKKS cause obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 67-70.	9.4	311
33	A recombination hotspot responsible for two inherited peripheral neuropathies is located near a mariner transposon-like element. Nature Genetics, 1996, 12, 288-297.	9.4	304
34	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
35	Molecular mechanism for duplication 17p11.2â€” the homologous recombination reciprocal of the Smith-Magenis microdeletion. Nature Genetics, 2000, 24, 84-87.	9.4	297
36	Two autosomal dominant neuropathies result from reciprocal DNA duplication/deletion of a region on chromosome 17. Human Molecular Genetics, 1994, 3, 223-228.	1.4	294

#	Article	IF	Citations
37	Mechanisms for recurrent and complex human genomic rearrangements. Current Opinion in Genetics and Development, 2012, 22, 211-220.	1.5	289
38	Multi-disciplinary clinical study of Smith-Magenis syndrome (deletion 17p11.2). , 1996, 62, 247-254.		285
39	Genomic Rearrangements and Gene Copy-Number Alterations as a Cause of Nervous System Disorders. Neuron, 2006, 52, 103-121.	3.8	284
40	Genotype/Phenotype Analysis of a Photoreceptor-Specific ATP-Binding Cassette Transporter Gene, ABCR, in Stargardt Disease. American Journal of Human Genetics, 1999, 64, 422-434.	2.6	277
41	Dejerineâ€“Sottas syndrome associated with point mutation in the peripheral myelin protein 22 (PMP22) gene. Nature Genetics, 1993, 5, 269-273.	9.4	274
42	Whole-Genome Sequencing for Optimized Patient Management. Science Translational Medicine, 2011, 03, 87re3.	5.8	272
43	Gene dosage is a mechanism for Charcot-Marie-Tooth disease type 1A. Nature Genetics, 1992, 1, 29-33.	9.4	270
44	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	1.5	261
45	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	3.8	258
46	Development and validation of a CGH microarray for clinical cytogenetic diagnosis. Genetics in Medicine, 2005, 7, 422-432.	1.1	241
47	Complex human chromosomal and genomic rearrangements. Trends in Genetics, 2009, 25, 298-307.	2.9	239
48	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	13.9	239
49	Somatic mosaicism: implications for disease and transmission genetics. Trends in Genetics, 2015, 31, 382-392.	2.9	234
50	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234
51	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
52	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	1.1	225
53	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	2.6	219
54	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211

#	Article	IF	Citations
55	Evidence for a recessive PMP22 point mutation in Charcotâ€“Marieâ€“Tooth disease type 1A. Nature Genetics, 1993, 5, 189-194.	9.4	208
56	TLR7 gain-of-function genetic variation causes human lupus. Nature, 2022, 605, 349-356.	13.7	208
57	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
58	Increased LIS1 expression affects human and mouse brain development. Nature Genetics, 2009, 41, 168-177.	9.4	199
59	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	13.5	189
60	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
61	Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics, 2011, 43, 1074-1081.	9.4	184
62	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
63	Myelin deficiencies in both the central and the peripheral nervous systems associated with aSOX10 mutation. Annals of Neurology, 1999, 46, 313-318.	2.8	181
64	Bardetâ€“Biedl syndrome is linked to DNA markers on chromosome 11 q and is genetically heterogeneous. Nature Genetics, 1994, 7, 108-112.	9.4	179
65	Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. Human Molecular Genetics, 2009, 18, 2188-2203.	1.4	165
66	Mus81 and converging forks limit the mutagenicity of replication fork breakage. Science, 2015, 349, 742-747.	6.0	162
67	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
68	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	2.6	160
69	Microarrayâ€based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. American Journal of Medical Genetics, Part A, 2007, 143A, 1679-1686.	0.7	158
70	Assessing structural variation in a personal genomeâ€”towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
71	The allelic spectrum of Charcotâ€“Marieâ€“Tooth disease in over 17,000 individuals with neuropathy. Molecular Genetics & Genomic Medicine, 2014, 2, 522-529.	0.6	151
72	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	2.6	150

#	Article	IF	Citations
73	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	2.6	148
74	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
75	Genomic Rearrangements Resulting in PLP1 Deletion Occur by Nonhomologous End Joining and Cause Different Dysmyelinating Phenotypes in Males and Females. American Journal of Human Genetics, 2002, 71, 838-853.	2.6	144
76	Genome Mosaicismâ€"One Human, Multiple Genomes. Science, 2013, 341, 358-359.	6.0	143
77	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57.	3.6	143
78	Diagnosis of CMT1A duplications and HNPP deletions by interphase FISH: Implications for testing in the cytogenetics laboratory. American Journal of Medical Genetics Part A, 1997, 69, 325-331.	2.4	141
79	Phenotypic Consequences of Copy Number Variation: Insights from Smith-Magenis and Potocki-Lupski Syndrome Mouse Models. PLoS Biology, 2010, 8, e1000543.	2.6	139
80	Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood, 2018, 132, 89-100.	0.6	139
81	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	3.9	136
82	Genomic disorders ten years on. Genome Medicine, 2009, 1, 42.	3.6	135
83	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
84	The 1.4-Mb CMT1A Duplication/HNPP Deletion Genomic Region Reveals Unique Genome Architectural Features and Provides Insights into the Recent Evolution of New Genes. Genome Research, 2001, 11, 1018-1033.	2.4	129
85	Novel mutations of MYO15A associated with profound deafness in consanguineous families and moderately severe hearing loss in a patient with Smith-Magenis syndrome. Human Genetics, 2001, 109, 535-541.	1.8	128
86	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	3.6	128
87	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
88	Cell cycle arrest in Era GTPase mutants: a potential growth rate-regulated checkpoint inEscherichia coli. Molecular Microbiology, 1998, 27, 739-750.	1.2	127
89	Novel genetic causes for cerebral visual impairment. European Journal of Human Genetics, 2016, 24, 660-665.	1.4	127
90	Proteolipid protein gene duplications causing Pelizaeus-Merzbacher disease: Molecular mechanism and phenotypic manifestations. Annals of Neurology, 1999, 45, 624-632.	2.8	126

#	Article	IF	Citations
91	Mechanisms for Nonrecurrent Genomic Rearrangements Associated with CMT1A or HNPP: Rare CNVs as a Cause for Missing Heritability. American Journal of Human Genetics, 2010, 86, 892-903.	2.6	125
92	Replicative mechanisms for CNV formation are error prone. Nature Genetics, 2013, 45, 1319-1326.	9.4	125
93	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
94	The Breakpoint Region of the Most Common Isochromosome, i(17q), in Human Neoplasia Is Characterized by a Complex Genomic Architecture with Large, Palindromic, Low-Copy Repeats. American Journal of Human Genetics, 2004, 74, 1-10.	2.6	122
95	Oral Curcumin Mitigates the Clinical and Neuropathologic Phenotype of the Trembler-J Mouse: A Potential Therapy for Inherited Neuropathy. American Journal of Human Genetics, 2007, 81, 438-453.	2.6	122
96	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	1.5	122
97	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	2.4	120
98	Structural variation mutagenesis of the human genome: Impact on disease and evolution. Environmental and Molecular Mutagenesis, 2015, 56, 419-436.	0.9	119
99	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117
100	Curcumin Treatment Abrogates Endoplasmic Reticulum Retention and Aggregation-Induced Apoptosis Associated with Neuropathy-Causing Myelin Protein Zeroâ€“Truncating Mutants. American Journal of Human Genetics, 2005, 77, 841-850.	2.6	115
101	DUF1220-Domain Copy Number Implicated in Human Brain-Size Pathology and Evolution. American Journal of Human Genetics, 2012, 91, 444-454.	2.6	113
102	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	1.4	112
103	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
104	The Centers for Mendelian Genomics: A new largeâ€scale initiative to identify the genes underlying rare Mendelian conditions. American Journal of Medical Genetics, Part A, 2012, 158A, 1523-1525.	0.7	110
105	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
106	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56.	2.4	110
107	The rod photoreceptor ATP-binding cassette transporter gene, ABCR, and retinal disease: from monogenic to multifactorial. Vision Research, 1999, 39, 2537-2544.	0.7	108
108	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	1.1	105

#	Article	IF	Citations
109	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
110	Parent of Origin, Mosaicism, and Recurrence Risk: Probabilistic Modeling Explains the Broken Symmetry of Transmission Genetics. American Journal of Human Genetics, 2014, 95, 345-359.	2.6	103
111	Modeling del(17)(p11.2p11.2) and dup(17)(p11.2p11.2) Contiguous Gene Syndromes by Chromosome Engineering in Mice: Phenotypic Consequences of Gene Dosage Imbalance. Molecular and Cellular Biology, 2003, 23, 3646-3655.	1.1	100
112	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
113	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	2.6	98
114	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	3.8	95
115	Delineation of the common critical region in Williams syndrome and clinical correlation of growth, heart defects, ethnicity, and parental origin. , 1998, 78, 82-89.		93
116	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	2.6	92
117	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	1.1	92
118	Pro-inflammation Associated with a Gain-of-Function Mutation (R284S) in the Innate Immune Sensor STING. Cell Reports, 2018, 23, 1112-1123.	2.9	92
119	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.	2.6	90
120	Unusual electrophysiological findings in X-linked dominant Charcot-Marie-Tooth disease. Muscle and Nerve, 2000, 23, 182-188.	1.0	89
121	Is the carboxyl-terminus of dystrophin required for membrane association? A novel, severe case of duchenne muscular dystrophy. Annals of Neurology, 1991, 30, 605-610.	2.8	88
122	DVL3 Alleles Resulting in a â^'1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	2.6	88
123	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
124	The Alu-Rich Genomic Architecture of SPAST Predisposes to Diverse and Functionally Distinct Disease-Associated CNV Alleles. American Journal of Human Genetics, 2014, 95, 143-161.	2.6	87
125	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	1.5	87
126	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86

#	Article	IF	Citations
127	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	1.8	85
128	Rapid molecular diagnostics of severe primary immunodeficiency determined by using targeted next-generation sequencing. Journal of Allergy and Clinical Immunology, 2016, 138, 1142-1151.e2.	1.5	85
129	Spastic paraplegia type 2 associated with axonal neuropathy and apparent PLP1 position effect. Annals of Neurology, 2006, 59, 398-403.	2.8	83
130	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	1.4	83
131	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778.	3.9	82
132	The human COX10 gene is disrupted during homologous recombination between the 24 kb proximal and distal CMT1A-REPs. Human Molecular Genetics, 1997, 6, 1595-1603.	1.4	81
133	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
134	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42.	0.7	80
135	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	1.5	80
136	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	2.6	79
137	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	3.6	78
138	Discordance of muscular dystrophy in monozygotic female twins: Evidence supporting asymmetric splitting of the inner cell mass in a manifesting carrier of Duchenne dystrophy. American Journal of Medical Genetics Part A, 1991, 40, 354-364.	2.4	76
139	Somatic mosaicism underlies X-linked acrogigantism syndrome in sporadic male subjects. Endocrine-Related Cancer, 2016, 23, 221-233.	1.6	75
140	Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Human Molecular Genetics, 2011, 20, 1975-1988.	1.4	74
141	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	1.1	74
142	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	2.4	74
143	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	2.6	74
144	Role of genomic architecture in PLP1 duplication causing Pelizaeus-Merzbacher disease. Human Molecular Genetics, 2006, 15, 2250-2265.	1.4	73

#	Article	IF	Citations
145	Reporting Genomic Sequencing Results to Ordering Clinicians. JAMA - Journal of the American Medical Association, 2013, 310, 365.	3.8	73
146	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	1.1	73
147	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
148	Observation and prediction of recurrent human translocations mediated by NAHR between nonhomologous chromosomes. Genome Research, 2011, 21, 33-46.	2.4	72
149	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	4.5	71
150	Mutations in ANKLE2, a ZIKA Virus Target, Disrupt an Asymmetric Cell Division Pathway in Drosophila Neuroblasts to Cause Microcephaly. Developmental Cell, 2019, 51, 713-729.e6.	3.1	71
151	Perturbations of BMP/TGF-Î² and VEGF/VEGFR signalling pathways in non-syndromic sporadic brain arteriovenous malformations (BAVM). Journal of Medical Genetics, 2018, 55, 675-684.	1.5	70
152	Approaches for identifying germ cell mutagens: Report of the 2013 IWGT workshop on germ cell assaysâ˜†. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2015, 783, 36-54.	0.9	69
153	DNA Rearrangements on Both Homologues of Chromosome 17 in a Mildly Delayed Individual with a Family History of Autosomal Dominant Carpal Tunnel Syndrome. American Journal of Human Genetics, 1999, 64, 471-478.	2.6	67
154	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129.	2.6	67
155	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	2.6	66
156	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
157	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
158	Rai1 duplication causes physical and behavioral phenotypes in a mouse model of dup(17)(p11.2p11.2). Journal of Clinical Investigation, 2006, 116, 3035-3041.	3.9	66
159	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	2.6	65
160	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207.	6.0	65
161	Hotspots of homologous recombination in the human genome: not all homologous sequences are equal. Genome Biology, 2004, 5, 242.	13.9	64
162	PacBio-LITS: a large-insert targeted sequencing method for characterization of human disease-associated chromosomal structural variations. BMC Genomics, 2015, 16, 214.	1.2	63

#	Article	IF	Citations
163	Deletions of recessive disease genes: CNV contribution to carrier states and disease-causing alleles. Genome Research, 2013, 23, 1383-1394.	2.4	62
164	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	2.6	62
165	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	3.7	62
166	EGR2 mutation R359W causes a spectrum of Dejerine-Sottas neuropathy. Neurogenetics, 2001, 3, 153-157.	0.7	60
167	Genomic disorders: A window into human gene and genome evolution. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1765-1771.	3.3	60
168	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558.	1.1	60
169	A potential founder variant inCARMIL2/RLTPRin three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. Molecular Genetics & Genomic Medicine, 2016, 4, 604-616.	0.6	59
170	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
171	T118M PMP22 mutation causes partial loss of function and HNPPâ€like neuropathy. Annals of Neurology, 2006, 59, 358-364.	2.8	58
172	Whole-exome sequencing identifies novel homozygous mutation inÂNPAS2 in family with nonobstructive azoospermia. Fertility and Sterility, 2015, 104, 286-291.	0.5	58
173	GNB5 Mutations Cause an Autosomal-Recessive Multisystem Syndrome with Sinus Bradycardia and Cognitive Disability. American Journal of Human Genetics, 2016, 99, 704-710.	2.6	58
174	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	1.1	58
175	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	1.1	58
176	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
177	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	2.6	57
178	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050.	1.5	57
179	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	2.6	57
180	The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. Human Genetics, 2018, 137, 553-567.	1.8	57

#	Article	IF	Citations
181	Conservation and evolution of the rpsU-dnaG-rpoD macromolecular synthesis operon in bacteria. Molecular Microbiology, 1993, 8, 343-355.	1.2	56
182	Myelin protein zero (MPZ) gene mutations in nonduplication type 1 Charcot-Marie-Tooth disease. , 1996, 7, 36-45.		56
183	Enriched rearing improves behavioral responses of an animal model for CNV-based autistic-like traits. Human Molecular Genetics, 2012, 21, 3083-3096.	1.4	56
184	Phenotypic expansion of TBX4 mutations to include acinar dysplasia of the lungs. American Journal of Medical Genetics, Part A, 2016, 170, 2440-2444.	0.7	56
185	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
186	Mutational and genotypeâ€“phenotype correlation analyses in 28 Polish patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1531-1541.	0.7	55
187	Allelic Mutations of KITLG, Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. American Journal of Human Genetics, 2015, 97, 647-660.	2.6	55
188	Digenic inheritance and Mendelian disease. Nature Genetics, 2012, 44, 1291-1292.	9.4	54
189	Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	4.5	54
190	Longitudinal studies of the duplication form of Charcot-Marie-Tooth polyneuropathy. , 1996, 19, 74-78.		53
191	Compensating for central nervous system dysmyelination: Females with a proteolipid protein gene duplication and sustained clinical improvement. Annals of Neurology, 2001, 50, 747-754.	2.8	53
192	Alu-specific microhomology-mediated deletion of the final exon of SPAST in three unrelated subjects with hereditary spastic paraplegia. Genetics in Medicine, 2011, 13, 582-592.	1.1	53
193	Mutations in COL27A1 cause Steel syndrome and suggest a founder mutation effect in the Puerto Rican population. European Journal of Human Genetics, 2015, 23, 342-346.	1.4	53
194	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	1.8	53
195	Characterization of molecular and cellular phenotypes associated with a heterozygous CNTNAP2 deletion using patient-derived hiPSC neural cells. NPJ Schizophrenia, 2015, 1, .	2.0	52
196	Germline PRKACA amplification causes variable phenotypes that may depend on the extent of the genomic defect: molecular mechanisms and clinical presentations. European Journal of Endocrinology, 2015, 172, 803-811.	1.9	52
197	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	1.4	52
198	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52

#	Article	IF	Citations
199	The Smith-Magenis syndrome [del(17)p11.2]: Clinical review and molecular advances. Mental Retardation and Developmental Disabilities Research Reviews, 1996, 2, 122-129.	3.5	51
200	Novel 9q34.11 gene deletions encompassing combinations of four Mendelian disease genes: STXBP1, SPTAN1, ENG, and TOR1A. Genetics in Medicine, 2012, 14, 868-876.	1.1	51
201	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	1.4	51
202	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
203	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220.	1.1	48
204	Molecular Genetics and Neuropathology of Charcot-Marie-Tooth Disease Type 1A. Brain Pathology, 1992, 2, 337-349.	2.1	47
205	Quantification by flow cytometry of chromosome-17 deletions in Smith-Magenis syndrome patients. Human Genetics, 1996, 98, 710-718.	1.8	47
206	Genomic Disorders: Recombination-Based Disease Resulting from Genome Architecture**Previously presented at the annual meeting of The American Society of Human Genetics, in Baltimore, on October 19, 2002.. American Journal of Human Genetics, 2003, 72, 246-252.	2.6	47
207	Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54.	3.6	47
208	Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippelâ€“Feil syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2795-2799.	0.7	47
209	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	5.8	47
210	RAI1 point mutations, CAG repeat variation, and SNP analysis in non-deletion Smithâ€“Magenis syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 2454-2463.	0.7	46
211	Incriminating genomic evidence. Nature, 2008, 455, 178-179.	13.7	46
212	TBX6 compound inheritance leads to congenital vertebral malformations in humans and mice. Human Molecular Genetics, 2019, 28, 539-547.	1.4	46
213	Absence of Heterozygosity Due to Template Switching during Replicative Rearrangements. American Journal of Human Genetics, 2015, 96, 555-564.	2.6	45
214	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	1.5	45
215	Aggressive tumor growth and clinical evolution in a patient with X-linked acro-gigantism syndrome. Endocrine, 2016, 51, 236-244.	1.1	45
216	Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. American Journal of Human Genetics, 2016, 98, 202-209.	2.6	45

#	Article	IF	Citations
217	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156.	2.6	44
218	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	1.4	44
219	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
220	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	3.6	43
221	Mechanism, Prevalence, and More Severe Neuropathy Phenotype of the Charcot-Marie-Tooth Type 1A Triplication. American Journal of Human Genetics, 2014, 94, 462-469.	2.6	42
222	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
223	A syndrome of short stature, microcephaly and speech delay is associated with duplications reciprocal to the common Sotos syndrome deletion. European Journal of Human Genetics, 2010, 18, 258-261.	1.4	41
224	A Human in Human Genetics. Cell, 2019, 177, 9-15.	13.5	41
225	Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	2.6	41
226	Charcot-Marie-Tooth Polyneuropathy: Duplication, Gene Dosage, and Genetic Heterogeneity. Pediatric Research, 1999, 45, 159-165.	1.1	41
227	Dosage Changes of a Segment at 17p13.1 Lead to Intellectual Disability and Microcephaly as a Result of Complex Genetic Interaction of Multiple Genes. American Journal of Human Genetics, 2014, 95, 565-578.	2.6	40
228	Passage Number is a Major Contributor to Genomic Structural Variations in Mouse iPSCs. Stem Cells, 2014, 32, 2657-2667.	1.4	40
229	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	1.8	40
230	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	9.4	40
231	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
232	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47.	1.5	40
233	A clinical and molecular study of mosaicism for trisomy 17. Human Genetics, 1996, 97, 69-72.	1.8	39
234	Reduced penetrance of craniofacial anomalies as a function of deletion size and genetic background in a chromosome engineered partial mouse model for Smithâ€“Magenis syndrome. Human Molecular Genetics, 2004, 13, 2613-2624.	1.4	39

#	Article	IF	Citations
235	Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19.	1.8	39
236	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	3.6	39
237	Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. Human Molecular Genetics, 2018, 27, 1913-1926.	1.4	39
238	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	2.6	38
239	New Polymorphic Short Tandem Repeats for PCR-based Charcot-Marie-Tooth Disease Type 1A Duplication Diagnosis. Clinical Chemistry, 2001, 47, 838-843.	1.5	37
240	Penetrance of Craniofacial Anomalies in Mouse Models of Smith-Magenis Syndrome Is Modified by Genomic Sequence Surrounding Rai1: Not All Null Alleles Are Alike. American Journal of Human Genetics, 2007, 80, 518-525.	2.6	37
241	Incidental copy-number variants identified by routine genome testing in a clinical population. Genetics in Medicine, 2013, 15, 45-54.	1.1	37
242	CHRNA7 triplication associated with cognitive impairment and neuropsychiatric phenotypes in a three-generation pedigree. European Journal of Human Genetics, 2014, 22, 1071-1076.	1.4	37
243	A visual and curatorial approach to clinical variant prioritization and disease gene discovery in genome-wide diagnostics. Genome Medicine, 2016, 8, 13.	3.6	37
244	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95.	3.6	37
245	Translation of SOX10 3' untranslated region causes a complex severe neurocristopathy by generation of a deleterious functional domain. Human Molecular Genetics, 2007, 16, 3037-3046.	1.4	36
246	A Duplication CNV That Conveys Traits Reciprocal to Metabolic Syndrome and Protects against Diet-Induced Obesity in Mice and Men. PLoS Genetics, 2012, 8, e1002713.	1.5	36
247	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	1.8	36
248	Mutations in PI3K110Î´ cause impaired natural killer cell function partially rescued by rapamycin treatment. Journal of Allergy and Clinical Immunology, 2018, 142, 605-617.e7.	1.5	36
249	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
250	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
251	Stable inheritance of the CMT1A DNA duplication in two patients with CMT1 and NF1. American Journal of Medical Genetics Part A, 1993, 45, 92-96.	2.4	35
252	Prenatal interphase FISH diagnosis ofPLP1 duplication associated with Pelizaeus-Merzbacher disease. Prenatal Diagnosis, 2001, 21, 1133-1136.	1.1	35

#	Article	IF	Citations
253	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
254	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131.	1.4	35
255	Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq1 1 0.784314 rgBT /Overloc	0.7	35
256	Charcot-Marie-Tooth Disease: A Gene-Dosage Effect. Hospital Practice (1995), 1997, 32, 83-122.	0.5	34
257	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 1246-1252.	1.4	34
258	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	0.7	34
259	Defining the breakpoints of proximal chromosome 14q rearrangements in nine patients using flow-sorted chromosomes. American Journal of Medical Genetics Part A, 2001, 102, 173-182.	2.4	33
260	New mutations and intellectual function. Nature Genetics, 2010, 42, 1036-1038.	9.4	33
261	Nonrecurrent 17p11.2p12 Rearrangement Events that Result in Two Concomitant Genomic Disorders: The PMP22-RAI1 Contiguous Gene Duplication Syndrome. American Journal of Human Genetics, 2015, 97, 691-707.	2.6	33
262	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
263	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. American Journal of Human Genetics, 2020, 106, 272-279.	2.6	33
264	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.	0.7	32
265	An evaluation of the draft human genome sequence. Nature Genetics, 2001, 29, 88-91.	9.4	32
266	Human subtelomeric copy number gains suggest a DNA replication mechanism for formation: beyond breakageâ€“fusionâ€“bridge for telomere stabilization. Human Genetics, 2012, 131, 1895-1910.	1.8	32
267	From genomic medicine to precision medicine: highlights of 2015. Genome Medicine, 2016, 8, 12.	3.6	32
268	Targeted Treatment of Individuals With Psychosis Carrying a Copy Number Variant Containing a Genomic Triplication of the Glycine Decarboxylase Gene. Biological Psychiatry, 2019, 86, 523-535.	0.7	32
269	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, .	4.2	31
270	NIPBL rearrangements in Cornelia de Lange syndrome: evidence for replicative mechanism and genotypeâ€“phenotype correlation. Genetics in Medicine, 2012, 14, 313-322.	1.1	30

#	Article	IF	Citations
271	Exonic duplication CNV of NDRG1 associated with autosomal-recessive HMSN-Lom/CMT4D. Genetics in Medicine, 2014, 16, 386-394.	1.1	30
272	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.	1.4	30
273	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	2.6	30
274	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
275	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
276	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	1.1	30
277	The Deep Genome Project. Genome Biology, 2020, 21, 18.	3.8	30
278	De novo proximal interstitial deletions of 14q: Cytogenetic and molecular investigations. American Journal of Medical Genetics Part A, 1994, 52, 44-50.	2.4	29
279	Multiplede novo MPZ (P0) point mutations in a sporadic Dejerine-Sottas case. , 1997, 10, 21-24.		29
280	Genitourinary Defects Associated with Genomic Deletions in 2p15 Encompassing OTX1. PLoS ONE, 2014, 9, e107028.	1.1	29
281	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	1.4	28
282	A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2018, 26, 1294-1305.	1.4	28
283	Exploring the utility of wholeâ€exome sequencing as a diagnostic tool in a child withÂatypical episodic muscle weakness. Clinical Genetics, 2013, 83, 457-461.	1.0	27
284	TBX6 missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	1.1	27
285	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	1.1	27
286	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. European Journal of Human Genetics, 2020, 28, 1243-1264.	1.4	27
287	Absence ofPMP22 coding region mutations in CMT1A duplication patients: Further evidence supporting gene dosage as a mechanism for charcot-marie-tooth disease type 1A. , 1996, 8, 362-365.		26
288	Recessive Charcot-Marie-Tooth disease. Annals of Neurology, 2000, 47, 6-8.	2.8	26

#	Article	IF	Citations
289	Clinical utility of whole-exome sequencing in rare diseases: Galactosialidosis. European Journal of Medical Genetics, 2014, 57, 339-344.	0.7	26
290	Dual molecular diagnosis contributes to atypical Praderâ€“Willi phenotype in monozygotic twins. American Journal of Medical Genetics, Part A, 2017, 173, 2451-2455.	0.7	26
291	Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946.	1.1	26
292	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	0.6	26
293	Harnessing genomics to identify environmental determinants of heritable disease. Mutation Research - Reviews in Mutation Research, 2013, 752, 6-9.	2.4	25
294	PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. Pediatric Neurology, 2016, 60, 83-87.	1.0	25
295	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
296	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
297	Clan genomics: From <scp>OMIM</scp> phenotypic traits to genes and biology. American Journal of Medical Genetics, Part A, 2021, 185, 3294-3313.	0.7	25
298	Smith-Magenis syndrome deletion: A case with equivocal cytogenetic findings resolved by fluorescence in situ hybridization. American Journal of Medical Genetics Part A, 1995, 58, 286-291.	2.4	24
299	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	1.5	24
300	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	1.8	24
301	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
302	Distinct patterns of complex rearrangements and a mutational signature of microhomeology are frequently observed in PLP1 copy number gain structural variants. Genome Medicine, 2019, 11, 80.	3.6	24
303	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	1.2	24
304	22q11.2q13 duplication including SOX10 causes sexâ€reversal and peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and Hirschsprung disease. American Journal of Medical Genetics, Part A, 2017, 173, 1066-1070.	0.7	23
305	Novel Combined Immune Deficiency and Radiation Sensitivity Blended Phenotype in an Adult with Biallelic Variations in ZAP70 and RNF168. Frontiers in Immunology, 2017, 8, 576.	2.2	23
306	Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. Journal of Medical Genetics, 2020, 57, 371-379.	1.5	23

#	Article	IF	Citations
307	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. Human Genetics, 2021, 140, 1011-1029.	1.8	23
308	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
309	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	1.1	22
310	An evolution revolution provides further revelation. BioEssays, 2007, 29, 1182-1184.	1.2	21
311	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. PLoS Genetics, 2015, 11, e1005686.	1.5	21
312	Exome sequencing reveals genetic architecture in patients with isolated or syndromic short stature. Journal of Genetics and Genomics, 2021, 48, 396-402.	1.7	21
313	Prenatal diagnosis of Charcot-Marie-Tooth disease type 1A by interphase fluorescencein situ hybridization. , 1999, 19, 446-449.		20
314	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105.	3.6	20
315	Efficient CNV breakpoint analysis reveals unexpected structural complexity and correlation of dosage-sensitive genes with clinical severity in genomic disorders. Human Molecular Genetics, 2017, 26, 1927-1941.	1.4	20
316	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
317	Missense mutations in the 3' end of the Escherichia coli dnaG gene do not abolish primase activity but do confer the chromosome-segregation-defective (par) phenotype. Microbiology (United Kingdom), 1997, 143, 585-594.	0.7	19
318	Prenatal ultrasonographic description and postnatal pathological findings in atelosteogenesis type 1. , 1998, 79, 392-395.		19
319	Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. European Journal of Human Genetics, 2014, 22, 1145-1148.	1.4	19
320	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1â€“q35.3 susceptibility locus identified by whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 73-78.	1.4	19
321	A large CRISPR-induced bystander mutation causes immune dysregulation. Communications Biology, 2019, 2, 70.	2.0	19
322	Genetic and molecular mechanism for distinct clinical phenotypes conveyed by allelic truncating mutations implicated in FBN1. Molecular Genetics & Genomic Medicine, 2020, 8, e1023.	0.6	19
323	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
324	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234.	1.1	18

#	Article	IF	Citations
325	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. Genetics in Medicine, 2016, 18, 443-451.	1.1	18
326	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	0.5	18
327	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	2.6	18
328	Novel Heterozygous Mutation in NFKB2 Is Associated With Early Onset CVID and a Functional Defect in NK Cells Complicated by Disseminated CMV Infection and Severe Nephrotic Syndrome. Frontiers in Pediatrics, 2019, 7, 303.	0.9	18
329	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
330	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	1.1	18
331	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
332	Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151.	0.7	17
333	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
334	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	1.8	17
335	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030.	2.6	17
336	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. Human Mutation, 2016, 37, 160-164.	1.1	16
337	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	1.4	16
338	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	0.7	16
339	NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531.	1.4	16
340	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	3.6	16
341	A novel homozygous <scp>SLC13A5</scp> wholeâ€gene deletion generated by <scp>Alu/Alu</scp>â€mediated rearrangement in an Iraqi family with epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 1972-1980.	0.7	16
342	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16

#	Article	IF	Citations
343	Dna ? DNA, and DNA ? RNA ? protein: Orchestration by a single complex operon. BioEssays, 1989, 10, 152-157.	1.2	15
344	Cerebral visual impairment and intellectual disability caused by PGAP1 variants. European Journal of Human Genetics, 2015, 23, 1689-1693.	1.4	15
345	CNV instability associated with DNA replication dynamics: evidence for replicative mechanisms in CNV mutagenesis. Human Molecular Genetics, 2015, 24, 1574-1583.	1.4	15
346	Clinical genomics: from a truly personal genome viewpoint. Human Genetics, 2016, 135, 591-601.	1.8	15
347	Prevalence of spinocerebellar ataxia 36 in a US population. Neurology: Genetics, 2017, 3, e174.	0.9	15
348	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.3	15
349	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	0.7	15
350	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168.	1.1	15
351	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	1.7	15
352	Biology in balance: human diploid genome integrity, gene dosage, and genomic medicine. Trends in Genetics, 2022, 38, 554-571.	2.9	15
353	Severe clinical phenotype due to an interstitial deletion of the short arm of chromosome 1: A brief review. , 1997, 71, 189-193.		14
354	Heterozygous CTNNB1 and TBX4 variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. Clinical Genetics, 2019, 96, 366-370.	1.0	14
355	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
356	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	1.3	14
357	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	0.7	14
358	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	2.8	14
359	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	1.1	14
360	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. American Journal of Medical Genetics, Part A, 2022, 188, 735-750.	0.7	14

#	Article	IF	Citations
361	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
362	Di George anomaly associated with ade novo Y;22 translocation resulting in monosomy del(22)(q11.2). American Journal of Medical Genetics Part A, 1991, 40, 196-198.	2.4	13
363	Platform comparison of detecting copy number variants with microarrays and whole-exome sequencing. Genomics Data, 2014, 2, 144-146.	1.3	13
364	From genes to genomes in the clinic. Genome Medicine, 2015, 7, 78.	3.6	13
365	First Case of CD40LG Deficiency in Ecuador, Diagnosed after Whole Exome Sequencing in a Patient with Severe Cutaneous Histoplasmosis. Frontiers in Pediatrics, 2017, 5, 17.	0.9	13
366	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. Molecular Genetics and Metabolism, 2018, 125, 302-304.	0.5	13
367	Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293.	0.7	13
368	Isolation and Characterization of Suppressors of Two Escherichia coli dnaG Mutations, dnaG2903 and parB. Genetics, 1997, 145, 867-875.	1.2	13
369	Opposing phenotypes in mice with Smithâ€“Magenis deletion and Potockâ€“Lupski duplication syndromes suggest gene dosage effects on fluid consumption behavior. American Journal of Medical Genetics, Part A, 2012, 158A, 2807-2814.	0.7	12
370	Duplications, deletions, and single-nucleotide variations: the complexity of genetic arithmetic. Genetics in Medicine, 2013, 15, 172-173.	1.1	12
371	Exome sequencing identifies a homozygous C5orf42 variant in a Turkish kindred with oralâ€facialâ€digital syndrome type VI. American Journal of Medical Genetics, Part A, 2015, 167, 2132-2137.	0.7	12
372	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
373	Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. Frontiers in Pediatrics, 2021, 9, 673957.	0.9	12
374	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	1.1	12
375	Cognitive Phenotypes and Genomic Copy Number Variations. JAMA - Journal of the American Medical Association, 2015, 313, 2029.	3.8	11
376	Phenotypic Expansion of Congenital Disorder of Glycosylation Due to SRD5A3 Null Mutation. JIMD Reports, 2015, 26, 7-12.	0.7	11
377	Two male sibs with severe micrognathia and a missense variant in MED12. European Journal of Medical Genetics, 2016, 59, 367-372.	0.7	11
378	Divergent Levels of Marker Chromosomes in an hiPSC-Based Model ofÂPsychosis. Stem Cell Reports, 2017, 8, 519-528.	2.3	11

#	Article	IF	Citations
379	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1015-1022.	0.7	11
380	2018 Victor A. McKusick Leadership Award: Molecular Mechanisms for Genomic and Chromosomal Rearrangements. American Journal of Human Genetics, 2019, 104, 391-406.	2.6	11
381	Ten years of Genome Medicine. Genome Medicine, 2019, 11, 7.	3.6	11
382	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	0.9	11
383	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	2.6	11
384	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
385	Whole-exome sequencing reveals an inherited R566X mutation of the epithelial sodium channel Î²-subunit in a case of early-onset phenotype of Liddle syndrome. Journal of Physical Education and Sports Management, 2016, 2, a001255.	0.5	10
386	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific C19orf12 isoform. Parkinsonism and Related Disorders, 2021, 82, 84-86.	1.1	10
387	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.0	10
388	Prenatal Diagnosis of Charcot-Marie-Tooth Disease Type 1A. Annals of the New York Academy of Sciences, 1999, 883, 457-459.	1.8	9
389	CHRNA7 Deletions are Enriched in Risperidone-Treated Children and Adolescents. Journal of Child and Adolescent Psychopharmacology, 2017, 27, 908-915.	0.7	9
390	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
391	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	1.1	9
392	Balancing between Adaptive and Maladaptive Cellular Stress Responses in Peripheral Neuropathy. Neuron, 2008, 57, 329-330.	3.8	8
393	Brain Copy Number Variants and Neuropsychiatric Traits. Biological Psychiatry, 2012, 72, 617-619.	0.7	8
394	CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473.	0.6	8
395	Chimeric transcripts resulting from complex duplications in chromosome Xq28. Human Genetics, 2016, 135, 253-256.	1.8	8
396	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753.	0.3	8

#	Article	IF	Citations
397	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
398	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	0.7	8
399	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
400	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	2.6	8
401	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053.	2.6	8
402	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
403	2012 highlights in translational 'omics. Genome Medicine, 2013, 5, 10.	3.6	7
404	SVachra: a tool to identify genomic structural variation in mate pair sequencing data containing inward and outward facing reads. BMC Genomics, 2017, 18, 691.	1.2	7
405	Xq26.3 Duplication in a Boy With Motor Delay and Low Muscle Tone Refines the X-Linked Acrogigantism Genetic Locus. Journal of the Endocrine Society, 2018, 2, 1100-1108.	0.1	7
406	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909.	0.7	7
407	Exome sequencing reveals a novel variant in NFX1 causing intracranial aneurysm in a Chinese family. Journal of NeuroInterventional Surgery, 2020, 12, 221-226.	2.0	7
408	Quantitative Assessment of Parental Somatic Mosaicism for Copyâ€Number Variant (CNV) Deletions. Current Protocols in Human Genetics, 2020, 106, e99.	3.5	7
409	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	0.7	7
410	Clinical presentation and evolution of Xiaâ€Gibbs syndrome due to p.Gly375ArgfsTer3 variant in a patient from DR Congo (Central Africa). American Journal of Medical Genetics, Part A, 2021, 185, 990-994.	0.7	7
411	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	1.7	7
412	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	1.0	7
413	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104.	1.7	7
414	Dominant Transmission Observed in Adolescents and Families With Orthostatic Intolerance. Pediatric Neurology, 2017, 66, 53-58.e5.	1.0	6

#	Article	IF	Citations
415	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurology: Genetics, 2021, 7, e589.	0.9	6
416	Risk of sudden cardiac death in <scp>EXOSC5</scp>â€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 2532-2540.	0.7	6
417	Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. Journal of Allergy and Clinical Immunology, 2022, 149, 758-766.	1.5	6
418	A Patient with Berardinelli-Seip Syndrome, Novel <i>AGPAT2</i> Splicesite Mutation and Concomitant Development of Non-diabetic Polyneuropathy. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 319-326.	0.4	6
419	Niacin therapy improves outcome and normalizes metabolic abnormalities in an NAXD-deficient patient. Brain, 2022, 145, e36-e40.	3.7	6
420	Settling the myelin protein zero question in CMT1B. Nature Genetics, 1995, 11, 119-120.	9.4	5
421	Animal models for human contiguous gene syndromes and other genomic disorders. Genetics and Molecular Biology, 2004, 27, 305-320.	0.6	5
422	Short stature and growth hormone deficiency in a subset of patients with <scp>Potockiâ€“Lupski</scp> syndrome: Expanding the phenotype of <scp>PTLS</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2077-2084.	0.7	5
423	Variants in FLRT3 and SLC35E2B identified using exome sequencing in seven high myopia families from Central Europe. Advances in Medical Sciences, 2021, 66, 192-198.	0.9	5
424	Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€“Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348.	1.1	5
425	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.0	5
426	Functional analysis of mutations in the transcription terminator T1 that suppress two dnaG alleles in Escherichia coli. Molecular Genetics and Genomics, 1995, 246, 729-733.	2.4	4
427	Duplication of thePMP22 gene in 17p partial trisomy patients with Charcot-Marie-Tooth type-1A neuropathy. Human Genetics, 1996, 97, 642-649.	1.8	4
428	Nonrecurrent PMP22-RAI1 contiguous gene deletions arise from replication-based mechanisms and result in Smithâ€“Magenis syndrome with evident peripheral neuropathy. Human Genetics, 2016, 135, 1161-1174.	1.8	4
429	Linking newborn severe combined immunodeficiency screening with targeted exome sequencing: A case report. Journal of Allergy and Clinical Immunology: in Practice, 2017, 5, 1442-1444.	2.0	4
430	Diagnosis of CMT1A duplications and HNPP deletions by interphase FISH: Implications for testing in the cytogenetics laboratory. American Journal of Medical Genetics Part A, 1997, 69, 325-331.	2.4	4
431	Novel RETREG1 (<scp>FAM134B</scp> founder allele is linked to <scp>HSAN2B</scp> and renal disease in a Turkish family. American Journal of Medical Genetics, Part A, 2022, 188, 2153-2161.	0.7	4
432	DNA rearrangements affecting dosage sensitive genes. Mental Retardation and Developmental Disabilities Research Reviews, 1996, 2, 139-146.	3.5	3

#	Article	IF	Citations
433	Regional localization of 10 mariner transposon-like ESTs by means of FISHâ€"evidence for a correlation with fragile sites. Mammalian Genome, 2001, 12, 326-328.	1.0	3
434	Objective measures of sleep disturbances in children with Potockiâ€"Lupski syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1982-1986.	0.7	3
435	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
436	Longitudinal studies of the duplication form of Charcot-Marie-Tooth polyneuropathy. , 1996, 19, 74.		3
437	Expanding the phenotypic and allelic spectrum of <scp>SMG8</scp>: Clinical observations reveal overlap with <scp>SMG9</scp>â€associated disease trait. American Journal of Medical Genetics, Part A, 2022, 188, 648-657.	0.7	3
438	Novel dominant and recessive variants in human ROBO1 cause distinct neurodevelopmental defects through different mechanisms. Human Molecular Genetics, 2022, 31, 2751-2765.	1.4	3
439	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	0.7	3
440	Short stature and combined immunodeficiency associated with mutations in RGS10. Science Signaling, 2021, 14, .	1.6	2
441	Proteolipid protein gene duplications causing Pelizaeus-Merzbacher disease: Molecular mechanism and phenotypic manifestations. , 1999, 45, 624.		2
442	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	2.8	2
443	Copy number analysis of the low-copy repeats at the primate NPHP1 locus by array comparative genomic hybridization. Genomics Data, 2016, 8, 106-109.	1.3	1
444	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Annals of Clinical and Translational Neurology, 2021, 8, 2052-2058.	1.7	1
445	The CMT1A Duplication. , 2006, , 3-17.		1
446	CRISPR/Cas9-induced gene conversion between ATAD3 paralogs. Human Genetics and Genomics Advances, 2022, 3, 100092.	1.0	1
447	Introduction of Arthur L. Beaudet, Harland Sanders Award Recipient. Genetics in Medicine, 2002, 4, 396-398.	1.1	0
448	Reply to InÃ¡cio et al. European Journal of Human Genetics, 2007, 15, 534-534.	1.4	0
449	Allan Award Introduction: Arthur L. Beaudet. American Journal of Human Genetics, 2008, 82, 1032-1033.	2.6	0
450	A Rare Novel Tyrosine Hydroxylase Gene Deletion in Parkinson Disease. Human Mutation, 2010, 31, v-v.	1.1	0

#	Article	IF	Citations
451	Front Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, i.	1.1	0
452	Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii.	1.1	0
453	Abstract 11800: Whole Exome Sequencing in a Large Pedigree With DCM Identifies a Novel Mutation in RBM20. Circulation, 2015, 132, .	1.6	0
454	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
455	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
456	MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, .	0.4	0
457	Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	1.1	0