James R Lupski

List of Publications by Year
in descending order

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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	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
2	Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. Journal of Allergy and Clinical Immunology, 2022, 149, 758-766.	1.5	6
3	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
4	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
5	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
6	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
7	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
8	CRISPR/Cas9-induced gene conversion between ATAD3 paralogs. Human Genetics and Genomics Advances, 2022, 3, 100092.	1.0	1
9	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
10	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
11	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. Npj Genomic Medicine, 2022, 7, 11.	1.7	7
12	<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
13	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
14	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
15	Niacin therapy improves outcome and normalizes metabolic abnormalities in an NAXD-deficient patient. Brain, 2022, 145, e36-e40.	3.7	6
16	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	1.1	11
17	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
18	Biology in balance: human diploid genome integrity, gene dosage, and genomic medicine. Trends in Genetics, 2022, 38, 554-571.	2.9	15

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19	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
20	TLR7 gain-of-function genetic variation causes human lupus. Nature, 2022, 605, 349-356.	13.7	208
21	MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, .	0.4	0
22	Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	1.1	0
23	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
24	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
25	Diagnostic yield and clinical impact of exome sequencing in early-onset scoliosis (EOS). Journal of Medical Genetics, 2021, 58, 41-47.	1.5	40
26	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
27	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
28	NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531.	1.4	16
29	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	0.6	26
30	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	2.8	14
31	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
32	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
33	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
34	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
35	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
36	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	3.6	16

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37	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
38	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurology: Genetics, 2021, 7, e589.	0.9	6
39	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
40	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
41	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
42	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
43	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, .	4.2	31
44	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
45	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
46	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
47	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
48	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	1.8	17
49	Short stature and combined immunodeficiency associated with mutations in RGS10. Science Signaling, 2021, 14, .	1.6	2
50	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
51	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
52	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
53	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
54	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16

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55	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
56	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	1.2	24
57	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Annals of Clinical and Translational Neurology, 2021, 8, 2052-2058.	1.7	1
58	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
59	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
60	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.0	5
61	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
62	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
63	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
64	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753.	0.3	8
65	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
66	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
67	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
68	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
69	TBX6 missense variants expand the mutational spectrum in a nonâ€Mendelian inheritance disease. Human Mutation, 2020, 41, 182-195.	1.1	27
70	Front Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, i.	1.1	0
71	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
72	Phenotypic expansion of POGZ â€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 6	0.7	35

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73	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
74	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
75	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
76	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
77	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
78	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
79	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
80	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
81	Clinical genomics and contextualizing genome variation in the diagnostic laboratory. Expert Review of Molecular Diagnostics, 2020, 20, 995-1002.	1.5	14
82	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
83	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	5.8	47
84	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	0.9	11
85	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
86	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
87	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
88	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
89	<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
90	Quantitative Assessment of Parental Somatic Mosaicism for Copyâ€Number Variant (CNV) Deletions. Current Protocols in Human Genetics, 2020, 106, e99.	3.5	7

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91	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
92	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207.	6.0	65
93	Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii.	1.1	0
94	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
95	The Deep Genome Project. Genome Biology, 2020, 21, 18.	3.8	30
96	Increased TBX6 gene dosages induce congenital cervical vertebral malformations in humans and mice. Journal of Medical Genetics, 2020, 57, 371-379.	1.5	23
97	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	1.7	15
98	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
99	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
100	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
101	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
102	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
103	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
104	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
105	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
106	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
107	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
108	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30

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109	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
110	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
111	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
112	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
113	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
114	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
115	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
116	A Human in Human Genetics. Cell, 2019, 177, 9-15.	13.5	41
117	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
118	Novel parent-of-origin-specific differentially methylated loci on chromosome 16. Clinical Epigenetics, 2019, 11, 60.	1.8	18
119	A large CRISPR-induced bystander mutation causes immune dysregulation. Communications Biology, 2019, 2, 70.	2.0	19
120	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
121	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
122	Ten years of Genome Medicine. Genome Medicine, 2019, 11, 7.	3.6	11
123	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
124	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
125	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
126	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	1.1	52

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127	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
128	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
129	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	1.4	44
130	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
131	TBX6 compound inheritance leads to congenital vertebral malformations in humans and mice. Human Molecular Genetics, 2019, 28, 539-547.	1.4	46
132	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
133	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
134	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	1.4	16
135	Genetic and mechanistic diversity in pediatric hemophagocytic lymphohistiocytosis. Blood, 2018, 132, 89-100.	0.6	139
136	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	1.4	52
137	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
138	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
139	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	0.7	34
140	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
141	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
142	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
143	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131.	1.4	35
144	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

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145	Mutations in the mitochondrial ribosomal protein MRPS22 lead to primary ovarian insufficiency. Human Molecular Genetics, 2018, 27, 1913-1926.	1.4	39
146	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
147	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
148	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
149	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	1.5	25
150	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
151	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
152	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
153	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
154	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
155	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
156	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
157	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
158	Predicting human genes susceptible to genomic instability associated with Alu/Alu-mediated rearrangements. Genome Research, 2018, 28, 1228-1242.	2.4	74
159	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
160	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
161	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
162	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

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