

# Shalini N Jhangiani

## List of Publications by Year in descending order

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Version: 2024-02-01

139  
papers

8,816  
citations

41258

49  
h-index

53109

85  
g-index

139  
all docs

139  
docs citations

139  
times ranked

17441  
citing authors

#	ARTICLE	IF	CITATIONS
1	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
2	Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 758-766.	1.5	6
3	Expanding the phenotypic and allelic spectrum of <i>SMG8</i> : Clinical observations reveal overlap with <i>SMG9</i> associated disease trait. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 648-657.	0.7	3
4	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 735-750.	0.7	14
5	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
6	Novel <i>RETREG1</i> ( <i>FAM134B</i> ) founder allele is linked to <i>HSAN2B</i> and renal disease in a Turkish family. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2153-2161.	0.7	4
7	Phenotypic and mutational spectrum of <i>ROR2</i> related Robinow syndrome. <i>Human Mutation</i> , 2022, 43, 900-918.	1.1	8
8	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	1.1	11
9	Biallelic Variants in the Ectonucleotidase <i>ENTPD1</i> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 92, 304-321.	2.8	2
10	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3593-3600.	0.7	16
11	Dominant mitochondrial membrane protein-associated neurodegeneration (MPAN) variants cluster within a specific <i>C19orf12</i> isoform. <i>Parkinsonism and Related Disorders</i> , 2021, 82, 84-86.	1.1	10
12	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
13	Neurodevelopmental disorder in an Egyptian family with a biallelic <i>ALKBH8</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	0.7	13
14	Perturbations of genes essential for Müllerian duct and Wolffian duct development in Mayer-Rokitansky-Küster-Hauser syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 337-345.	2.6	41
15	Exome sequencing reveals predominantly de novo variants in disorders with intellectual disability (ID) in the founder population of Finland. <i>Human Genetics</i> , 2021, 140, 1011-1029.	1.8	23
16	A novel homozygous <i>SLC13A5</i> whole-gene deletion generated by <i>Alu/Alu</i> mediated rearrangement in an Iraqi family with epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1972-1980.	0.7	16
17	Biallelic Pathogenic Variants in <i>TNNT3</i> Associated With Congenital Myopathy. <i>Neurology: Genetics</i> , 2021, 7, e589.	0.9	6
18	Two novel biallelic <i>KDEL2</i> missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2241-2249.	0.7	7

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19	Biallelic and monoallelic variants in <i>PLXNA1</i> are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	1.1	22
20	Risk of sudden cardiac death in <i>EXOSC5</i> -related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2532-2540.	0.7	6
21	<i>IFIH1</i> loss-of-function variants contribute to very early-onset inflammatory bowel disease. <i>Human Genetics</i> , 2021, 140, 1299-1312.	1.8	17
22	Exome variant discrepancies due to reference-genome differences. <i>American Journal of Human Genetics</i> , 2021, 108, 1239-1250.	2.6	36
23	Biallelic loss-of-function variants in the splicing regulator <i>NSRP1</i> cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 2455-2460.	1.1	9
24	Exome sequencing in children with clinically suspected maturity-onset diabetes of the young. <i>Pediatric Diabetes</i> , 2021, 22, 960-968.	1.2	6
25	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 365.	1.2	24
26	<i>COPB2</i> loss of function causes a coatopathy with osteoporosis and developmental delay. <i>American Journal of Human Genetics</i> , 2021, 108, 1710-1724.	2.6	18
27	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic <i>EMD</i> splice variant. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2052-2058.	1.7	1
28	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. <i>American Journal of Human Genetics</i> , 2021, 108, 1981-2005.	2.6	38
29	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in <i>RBM20</i> . <i>Acta Cardiologica</i> , 2020, 75, 748-753.	0.3	8
30	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. <i>Human Mutation</i> , 2020, 41, 487-501.	1.1	58
31	Deficiencies in vesicular transport mediated by <i>TRAPPC4</i> are associated with severe syndromic intellectual disability. <i>Brain</i> , 2020, 143, 112-130.	3.7	33
32	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. <i>Human Mutation</i> , 2020, 41, 641-654.	1.1	27
33	Biallelic in-frame deletion in <i>TRAPPC4</i> in a family with developmental delay and cerebellar atrophy. <i>Brain</i> , 2020, 143, e83-e83.	3.7	8
34	Congenital diaphragmatic hernia as a prominent feature of a <i>SPECC1L</i> -related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2919-2925.	0.7	8
35	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. <i>Genetics in Medicine</i> , 2020, 22, 1768-1776.	1.1	30
36	Phenotypic expansion in <i>KIF1A</i> -related dominant disorders: A description of novel variants and review of published cases. <i>Human Mutation</i> , 2020, 41, 2094-2104.	1.1	8

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37	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. <i>Neurology: Genetics</i> , 2020, 6, e498.	0.9	11
38	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1387-1399.	0.7	14
39	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. <i>Science</i> , 2020, 369, 202-207.	6.0	65
40	Biallelic <i>GRM7</i> variants cause epilepsy, microcephaly, and cerebral atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 610-627.	1.7	15
41	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. <i>Frontiers in Pediatrics</i> , 2019, 7, 303.	0.9	18
42	Heterozygous <i>CTNNB1</i> and <i>TBX4</i> variants in a patient with abnormal lung growth, pulmonary hypertension, microcephaly, and spasticity. <i>Clinical Genetics</i> , 2019, 96, 366-370.	1.0	14
43	Biallelic <i>CACNA2D2</i> variants in epileptic encephalopathy and cerebellar atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1395-1406.	1.7	20
44	The Genomics of Arthrogyrosis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. <i>American Journal of Human Genetics</i> , 2019, 105, 132-150.	2.6	74
45	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	2.6	56
46	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. <i>Journal of Experimental Medicine</i> , 2019, 216, 2778-2799.	4.2	132
47	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 1048-1056.	2.6	30
48	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
49	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	2.6	24
50	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 3049-3067.	1.8	53
51	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. <i>Genome Medicine</i> , 2019, 11, 25.	3.6	22
52	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
53	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27
54	Insights into genetics, human biology and disease gleaned from family based genomic studies. <i>Genetics in Medicine</i> , 2019, 21, 798-812.	1.1	161

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55	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. <i>American Journal of Human Genetics</i> , 2019, 104, 213-228.	2.6	90
56	Genetic architecture of laterality defects revealed by whole exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 563-573.	1.4	44
57	A Patient with Berardinelli-Seip Syndrome, Novel <i>AGPAT2</i> Splicesite Mutation and Concomitant Development of Non-diabetic Polyneuropathy. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2019, 11, 319-326.	0.4	6
58	Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. <i>Blood</i> , 2019, 134, 83-83.	0.6	0
59	A biallelic <i>ANTXR1</i> variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1015-1022.	0.7	11
60	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. <i>Nature Communications</i> , 2018, 9, 859.	5.8	126
61	The role of <i>FREM2</i> and <i>FRAS1</i> in the development of congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2018, 27, 2064-2075.	1.4	16
62	Biallelic variants in <i>KIF14</i> cause intellectual disability with microcephaly. <i>European Journal of Human Genetics</i> , 2018, 26, 330-339.	1.4	52
63	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 27-43.	2.6	88
64	Mutations in <i>PI3K110<math>\beta</math></i> cause impaired natural killer cell function partially rescued by rapamycin treatment. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 605-617.e7.	1.5	36
65	Phenotypic expansion illuminates multilocus pathogenic variation. <i>Genetics in Medicine</i> , 2018, 20, 1528-1537.	1.1	104
66	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. <i>European Journal of Human Genetics</i> , 2018, 26, 1121-1131.	1.4	35
67	Mutations in the mitochondrial ribosomal protein <i>MRPS22</i> lead to primary ovarian insufficiency. <i>Human Molecular Genetics</i> , 2018, 27, 1913-1926.	1.4	39
68	Identification of a pathogenic <i>PMP2</i> variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 302-304.	0.5	13
69	Mutation in the intracellular chloride channel <i>CLCC1</i> associated with autosomal recessive retinitis pigmentosa. <i>PLoS Genetics</i> , 2018, 14, e1007504.	1.5	25
70	Heterozygous Truncating Variants in <i>POMP</i> Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. <i>American Journal of Human Genetics</i> , 2018, 102, 1126-1142.	2.6	128
71	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. <i>Journal of Pediatric Genetics</i> , 2018, 07, 164-173.	0.3	15
72	Identification of likely pathogenic and known variants in <i>TSPEAR</i> , <i>LAMB3</i> , <i>BCOR</i> , and <i>WNT10A</i> in four Turkish families with tooth agenesis. <i>Human Genetics</i> , 2018, 137, 689-703.	1.8	24

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73	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. <i>American Journal of Human Genetics</i> , 2018, 103, 171-187.	2.6	160
74	A novel NAA10 variant with impaired acetyltransferase activity causes developmental delay, intellectual disability, and hypertrophic cardiomyopathy. <i>European Journal of Human Genetics</i> , 2018, 26, 1294-1305.	1.4	28
75	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1897-1909.	0.7	7
76	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. <i>Nucleic Acids Research</i> , 2017, 45, gkw1237.	6.5	98
77	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. <i>Genetics in Medicine</i> , 2017, 19, 13-19.	1.1	74
78	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. <i>Cell</i> , 2017, 168, 830-842.e7.	13.5	66
79	Dual molecular diagnosis contributes to atypical Prader-Willi phenotype in monozygotic twins. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2451-2455.	0.7	26
80	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a <i>TUBB3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a000984.	0.5	18
81	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
82	Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 1246-1252.	1.4	34
83	Analyses of SLC13A5 -epilepsy patients reveal perturbations of TCA cycle. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 314-319.	0.5	48
84	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. <i>American Journal of Human Genetics</i> , 2017, 101, 149-156.	2.6	44
85	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. <i>Genetics in Medicine</i> , 2017, 19, 412-420.	1.1	73
86	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1-q35.3 susceptibility locus identified by whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2017, 25, 73-78.	1.4	19
87	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 232-245.	1.5	261
88	First Case of CD40LG Deficiency in Ecuador, Diagnosed after Whole Exome Sequencing in a Patient with Severe Cutaneous Histoplasmosis. <i>Frontiers in Pediatrics</i> , 2017, 5, 17.	0.9	13
89	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. <i>Genome Medicine</i> , 2017, 9, 95.	3.6	37
90	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. <i>Genome Medicine</i> , 2016, 8, 106.	3.6	43

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91	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. <i>Human Genetics</i> , 2016, 135, 569-586.	1.8	85
92	Association of <i>MTOR</i> Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. <i>JAMA Neurology</i> , 2016, 73, 836.	4.5	234
93	CAV3 mutation in a patient with transient hyperCKemia and myalgia. <i>Neurologia I Neurochirurgia Polska</i> , 2016, 50, 468-473.	0.6	8
94	Bi-allelic Mutations in <i>PKD1L1</i> Are Associated with Laterality Defects in Humans. <i>American Journal of Human Genetics</i> , 2016, 99, 886-893.	2.6	57
95	Phenotypic expansion of <i>TBX4</i> mutations to include acinar dysplasia of the lungs. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2440-2444.	0.7	56
96	Recurrent De Novo and Biallelic Variation of <i>ATAD3A</i> , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	2.6	146
97	Identification of a <i>RAI1</i> -associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. <i>Genome Medicine</i> , 2016, 8, 105.	3.6	20
98	PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. <i>Pediatric Neurology</i> , 2016, 60, 83-87.	1.0	25
99	<i>POGZ</i> truncating alleles cause syndromic intellectual disability. <i>Genome Medicine</i> , 2016, 8, 3.	3.6	78
100	Two male sibs with severe micrognathia and a missense variant in <i>MED12</i> . <i>European Journal of Medical Genetics</i> , 2016, 59, 367-372.	0.7	11
101	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. <i>Human Mutation</i> , 2016, 37, 804-811.	1.1	92
102	Novel mutations in <i>LRP6</i> highlight the role of WNT signaling in tooth agenesis. <i>Genetics in Medicine</i> , 2016, 18, 1158-1162.	1.1	58
103	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. <i>Human Mutation</i> , 2016, 37, 231-234.	1.1	18
104	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic <i>TANGO2</i> Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	2.6	98
105	<i>DVL3</i> Alleles Resulting in a +1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2016, 98, 553-561.	2.6	88
106	Monoallelic and Biallelic Variants in <i>EMC1</i> Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 562-570.	2.6	66
107	Whole-Exome Sequencing in Familial Parkinson Disease. <i>JAMA Neurology</i> , 2016, 73, 68.	4.5	71
108	The role of combined SNV and CNV burden in patients with distal symmetric polyneuropathy. <i>Genetics in Medicine</i> , 2016, 18, 443-451.	1.1	18

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109	Novel genetic causes for cerebral visual impairment. <i>European Journal of Human Genetics</i> , 2016, 24, 660-665.	1.4	127
110	Secondary findings and carrier test frequencies in a large multiethnic sample. <i>Genome Medicine</i> , 2015, 7, 54.	3.6	47
111	Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippel-Feil syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2795-2799.	0.7	47
112	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 904-913.	2.6	65
113	Cerebral visual impairment and intellectual disability caused by PGAP1 variants. <i>European Journal of Human Genetics</i> , 2015, 23, 1689-1693.	1.4	15
114	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. <i>Cell Reports</i> , 2015, 12, 1169-1183.	2.9	211
115	A Massive Expansion of Effector Genes Underlies Gall-Formation in the Wheat Pest <i>Mayetiola destructor</i> . <i>Current Biology</i> , 2015, 25, 613-620.	1.8	171
116	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. <i>Journal of the National Cancer Institute</i> , 2015, 107, 384.	3.0	172
117	Targeted Sequencing in Chromosome 17q Linkage Region Identifies Familial Glioma Candidates in the Gliogene Consortium. <i>Scientific Reports</i> , 2015, 5, 8278.	1.6	22
118	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. <i>American Journal of Human Genetics</i> , 2015, 97, 199-215.	2.6	574
119	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. <i>Nature Genetics</i> , 2015, 47, 654-660.	9.4	302
120	Exome sequencing identifies a homozygous <i>C5orf42</i> variant in a Turkish kindred with oral-facial-digital syndrome type VI. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2132-2137.	0.7	12
121	New Mutations in the <i>RAB28</i> Gene in 2 Spanish Families With Cone-Rod Dystrophy. <i>JAMA Ophthalmology</i> , 2015, 133, 133.	1.4	28
122	Whole-exome sequencing identifies novel homozygous mutation in <i>NPAS2</i> in family with nonobstructive azoospermia. <i>Fertility and Sterility</i> , 2015, 104, 286-291.	0.5	58
123	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 612-622.	2.6	110
124	Allelic Mutations of <i>KITLG</i> , Encoding KIT Ligand, Cause Asymmetric and Unilateral Hearing Loss and Waardenburg Syndrome Type 2. <i>American Journal of Human Genetics</i> , 2015, 97, 647-660.	2.6	55
125	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. <i>Neuron</i> , 2015, 88, 499-513.	3.8	258
126	Phenotypic Expansion of Congenital Disorder of Glycosylation Due to <i>SRD5A3</i> Null Mutation. <i>JIMD Reports</i> , 2015, 26, 7-12.	0.7	11



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127	Hemichordate genomes and deuterostome origins. <i>Nature</i> , 2015, 527, 459-465.	13.7	217
128	Mutations in COL27A1 cause Steel syndrome and suggest a founder mutation effect in the Puerto Rican population. <i>European Journal of Human Genetics</i> , 2015, 23, 342-346.	1.4	53
129	Abstract 11800: Whole Exome Sequencing in a Large Pedigree With DCM Identifies a Novel Mutation in RBM20. <i>Circulation</i> , 2015, 132, .	1.6	0
130	Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. <i>European Journal of Human Genetics</i> , 2014, 22, 1145-1148.	1.4	19
131	The First Myriapod Genome Sequence Reveals Conservative Arthropod Gene Content and Genome Organisation in the Centipede <i>Strigamia maritima</i> . <i>PLoS Biology</i> , 2014, 12, e1002005.	2.6	221
132	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. <i>PLoS Genetics</i> , 2014, 10, e1004258.	1.5	122
133	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. <i>Cell</i> , 2014, 157, 636-650.	13.5	189
134	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 303-309.	2.6	125
135	The sheep genome illuminates biology of the rumen and lipid metabolism. <i>Science</i> , 2014, 344, 1168-1173.	6.0	436
136	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. <i>American Journal of Human Genetics</i> , 2014, 94, 915-923.	2.6	79
137	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. <i>American Journal of Human Genetics</i> , 2014, 95, 96-107.	2.6	148
138	Mutations in <i>VRK1</i> Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. <i>JAMA Neurology</i> , 2013, 70, 1491-8.	4.5	54
139	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. <i>Genome Medicine</i> , 2013, 5, 57.	3.6	143