

Vandana Shashi

List of Publications by Year in descending order

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Version: 2024-02-01

72
papers

3,741
citations

159585

30
h-index

144013

57
g-index

75
all docs

75
docs citations

75
times ranked

7003
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical application of exome sequencing in undiagnosed genetic conditions. <i>Journal of Medical Genetics</i> , 2012, 49, 353-361.	3.2	377
2	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. <i>Genetics in Medicine</i> , 2015, 17, 774-781.	2.4	284
3	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	27.0	261
4	The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. <i>Genetics in Medicine</i> , 2014, 16, 176-182.	2.4	239
5	Cognitive Decline Preceding the Onset of Psychosis in Patients With 22q11.2 Deletion Syndrome. <i>JAMA Psychiatry</i> , 2015, 72, 377.	11.0	196
6	Quinidine in the treatment of KCNT1-positive epilepsies. <i>Annals of Neurology</i> , 2015, 78, 995-999.	5.3	184
7	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	6.2	142
8	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. <i>American Journal of Human Genetics</i> , 2015, 96, 462-473.	6.2	124
9	Not the End of the Odyssey: Parental Perceptions of Whole Exome Sequencing (WES) in Pediatric Undiagnosed Disorders. <i>Journal of Genetic Counseling</i> , 2016, 25, 1019-1031.	1.6	91
10	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. <i>Nature Medicine</i> , 2020, 26, 1912-1918.	30.7	90
11	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. <i>Molecular Psychiatry</i> , 2021, 26, 4496-4510.	7.9	87
12	Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	7.8	86
13	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. <i>American Journal of Human Genetics</i> , 2016, 99, 711-719.	6.2	81
14	Infantile spasms and encephalopathy without preceding neonatal seizures caused by KCNQ2 R198Q, a gain-of-function variant. <i>Epilepsia</i> , 2017, 58, e10-e15.	5.1	81
15	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	6.2	69
16	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	6.2	68
17	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. <i>Genetics in Medicine</i> , 2019, 21, 1585-1593.	2.4	67
18	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	2.4	60

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19	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
20	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. <i>Human Molecular Genetics</i> , 2018, 27, 2454-2465.	2.9	54
21	A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 71.	2.7	53
22	Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?. <i>Journal of Genetic Counseling</i> , 2018, 27, 935-946.	1.6	49
23	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. <i>Schizophrenia Bulletin</i> , 2017, 43, 1079-1089.	4.3	47
24	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. <i>American Journal of Human Genetics</i> , 2019, 105, 283-301.	6.2	46
25	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. <i>Genetics in Medicine</i> , 2018, 20, 464-469.	2.4	42
26	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. <i>Journal of Genetic Counseling</i> , 2019, 28, 1107-1118.	1.6	42
27	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	1.3	42
28	Evidence of gray matter reduction and dysfunction in chromosome 22q11.2 deletion syndrome. <i>Psychiatry Research - Neuroimaging</i> , 2010, 181, 1-8.	1.8	39
29	The importance of dynamic re-analysis in diagnostic whole exome sequencing. <i>Journal of Medical Genetics</i> , 2017, 54, 155-156.	3.2	38
30	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	6.2	35
31	Epilepsy in <i>KCNH1</i> -related syndromes. <i>Epileptic Disorders</i> , 2016, 18, 123-136.	1.3	34
32	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 640-657.	6.2	31
33	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.	6.2	30
34	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. <i>Genetics in Medicine</i> , 2020, 22, 1269-1275.	2.4	30
35	Further evidence for the involvement of <i>EFL1</i> in a Shwachman-Diamond-like syndrome and expansion of the phenotypic features. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003046.	1.2	29
36	Exome sequencing results in successful riboflavin treatment of a rapidly progressive neurological condition. <i>Journal of Physical Education and Sports Management</i> , 2015, 1, a000257.	1.2	24

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37	Characteristics of undiagnosed diseases network applicants: implications for referring providers. <i>BMC Health Services Research</i> , 2018, 18, 652.	2.2	23
38	Phenotypic expansion of <i>KMT2D</i> -related disorder: Beyond Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1053-1065.	1.2	23
39	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. <i>Genetics in Medicine</i> , 2021, 23, 352-362.	2.4	23
40	COMT and anxiety and cognition in children with chromosome 22q11.2 deletion syndrome. <i>Psychiatry Research</i> , 2010, 178, 433-436.	3.3	20
41	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	2.4	20
42	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. <i>Schizophrenia Research</i> , 2019, 204, 320-325.	2.0	19
43	Heterozygous variants in <i>MYBPC1</i> are associated with an expanded neuromuscular phenotype beyond arthrogyposis. <i>Human Mutation</i> , 2019, 40, 1115-1126.	2.5	19
44	D-DEM ⁺ , a distinct phenotype caused by <i>ATP1A3</i> mutations. <i>Neurology: Genetics</i> , 2020, 6, e466.	1.9	18
45	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	2.4	18
46	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	3.8	18
47	Altered Development of the Dorsolateral Prefrontal Cortex in Chromosome 22q11.2 Deletion Syndrome: An In Vivo Proton Spectroscopy Study. <i>Biological Psychiatry</i> , 2012, 72, 684-691.	1.3	17
48	Increased corpus callosum volume in children with chromosome 22q11.2 deletion syndrome is associated with neurocognitive deficits and genetic polymorphisms. <i>European Journal of Human Genetics</i> , 2012, 20, 1051-1057.	2.8	17
49	Rare deleterious <i>de novo</i> missense variants in <i>Rnf2/Ring2</i> are associated with a neurodevelopmental disorder with unique clinical features. <i>Human Molecular Genetics</i> , 2021, 30, 1283-1292.	2.9	17
50	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1397.	1.2	16
51	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. <i>Genetics in Medicine</i> , 2021, 23, 1922-1932.	2.4	16
52	De novo variants in <i>TCF7L2</i> are associated with a syndromic neurodevelopmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2384-2390.	1.2	13
53	A normative chart for cognitive development in a genetically selected population. <i>Neuropsychopharmacology</i> , 2022, 47, 1379-1386.	5.4	12
54	Detection of a mosaic <i>CDKL5</i> deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1665.	1.2	11

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55	A pathogenic variant in the <i>SETBP1</i> hotspot results in a form of Schinzel-Giedion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1947-1951.	1.2	11
56	Communication of Psychiatric Risk in 22q11.2 Deletion Syndrome: A Pilot Project. <i>Journal of Genetic Counseling</i> , 2016, 25, 6-17.	1.6	9
57	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. <i>European Journal of Human Genetics</i> , 2021, 29, 271-279.	2.8	8
58	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. <i>American Journal of Human Genetics</i> , 2022, 109, 518-532.	6.2	8
59	The genome empowerment scale: An assessment of parental empowerment in families with undiagnosed disease. <i>Clinical Genetics</i> , 2019, 96, 521-531.	2.0	7
60	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. <i>Human Molecular Genetics</i> , 2019, 28, 3724-3733.	2.9	7
61	Bi-allelic <i>KARS1</i> pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. <i>Human Mutation</i> , 2021, 42, 745-761.	2.5	7
62	Frontal Hypoactivation During a Working Memory Task in Children With 22q11 Deletion Syndrome. <i>Journal of Child Neurology</i> , 2017, 32, 94-99.	1.4	6
63	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 361-372.	6.2	6
64	The microRNA processor <i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. <i>Human Molecular Genetics</i> , 2022, 31, 2934-2950.	2.9	6
65	Expanding the phenotypic spectrum of ARCN1-related syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1227-1237.	2.4	5
66	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	2.4	4
67	Epileptic encephalopathy with features of rapid-onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with <i>ATP1A3</i> mutation. <i>Epileptic Disorders</i> , 2020, 22, 103-109.	1.3	4
68	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. <i>Journal of Genetic Counseling</i> , 2022, 31, 59-70.	1.6	3
69	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. <i>Brain</i> , 2022, 145, 3383-3390.	7.6	3
70	Epilepsy in trisomy 7 mosaicism: A case report and literature review. <i>Journal of Pediatric Neurology</i> , 2015, 09, 063-068.	0.2	2
71	Completing the puzzle: The search for pieces in the understanding of psychosis risk in 22q11.2 deletion syndrome. <i>Schizophrenia Research</i> , 2017, 188, 33-34.	2.0	1
72	Hypogyrfication and its association with cognitive impairment in children with 22q11.2 deletion Syndrome: A preliminary report. <i>Psychiatry Research - Neuroimaging</i> , 2019, 285, 47-50.	1.8	0