Vandana Shashi

List of Publications by Year
in descending order

72 papers	3,741 citations	159525 30 h-index	143943 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. Journal of Medical Genetics, 2012, 49, 353-361.	1.5	377
2	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	1.1	284
3	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	13.9	261
4	The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. Genetics in Medicine, 2014, 16, 176-182.	1.1	239
5	Cognitive Decline Preceding the Onset of Psychosis in Patients With 22q11.2 Deletion Syndrome. JAMA Psychiatry, 2015, 72, 377.	6.0	196
6	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	2.8	184
7	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
8	De Novo Mutations in NALCN Cause a Syndrome Characterized by Congenital Contractures of the Limbs and Face, Hypotonia, and Developmental Delay. American Journal of Human Genetics, 2015, 96, 462-473.	2.6	124
9	Not the End of the Odyssey: Parental Perceptions of Whole Exome Sequencing (WES) in Pediatric Undiagnosed Disorders. Journal of Genetic Counseling, 2016, 25, 1019-1031.	0.9	91
10	Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome. Nature Medicine, 2020, 26, 1912-1918.	15.2	90
11	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
12	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
13	De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American Journal of Human Genetics, 2016, 99, 711-719.	2.6	81
14	Infantile spasms and encephalopathy without preceding neonatal seizures caused by KCNQ2 R198Q, a gainâ€ofâ€function variant. Epilepsia, 2017, 58, e10-e15.	2.6	81
15	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
16	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68
17	ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis. Genetics in Medicine, 2019, 21, 1585-1593.	1.1	67
18	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	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. American Journal of Human Genetics, 2019, 104, 164-178.	2.6	59
20	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	1.4	54
21	A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. Orphanet Journal of Rare Diseases, 2017, 12, 71.	1.2	53
22	Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?. Journal of Genetic Counseling, 2018, 27, 935-946.	0.9	49
23	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. Schizophrenia Bulletin, 2017, 43, 1079-1089.	2.3	47
24	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
25	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	1.1	42
26	Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. Journal of Genetic Counseling, 2019, 28, 1107-1118.	0.9	42
27	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
28	Evidence of gray matter reduction and dysfunction in chromosome 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2010, 181, 1-8.	0.9	39
29	The importance of dynamic re-analysis in diagnostic whole exome sequencing. Journal of Medical Genetics, 2017, 54, 155-156.	1.5	38
30	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
31	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	0.7	34
32	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	2.6	31
33	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
34	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genetics in Medicine, 2020, 22, 1269-1275.	1.1	30
35	Further evidence for the involvement of EFL1 in a Shwachmanâ€“Diamond-like syndrome and expansion of the phenotypic features. Journal of Physical Education and Sports Management, 2018, 4, a003046.	0.5	29
36	Exome sequencing results in successful riboflavin treatment of a rapidly progressive neurological condition. Journal of Physical Education and Sports Management, 2015, 1, a000257.	0.5	24

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37	Characteristics of undiagnosed diseases network applicants: implications for referring providers. BMC Health Services Research, 2018, 18, 652.	0.9	23
38	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	0.7	23
39	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	1.1	23
40	COMT and anxiety and cognition in children with chromosome 22q11.2 deletion syndrome. Psychiatry Research, 2010, 178, 433-436.	1.7	20
41	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
42	Attention deficit hyperactivity disorder symptoms as antecedents of later psychotic outcomes in 22q11.2 deletion syndrome. Schizophrenia Research, 2019, 204, 320-325.	1.1	19
43	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
44	D-DEMÃ˜, a distinct phenotype caused by ATP1A3 mutations. Neurology: Genetics, 2020, 6, e466.	0.9	18
45	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
46	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	1.8	18
47	Altered Development of the Dorsolateral Prefrontal Cortex in Chromosome 22q11.2 Deletion Syndrome: An In Vivo Proton Spectroscopy Study. Biological Psychiatry, 2012, 72, 684-691.	0.7	17
48	Increased corpus callosum volume in children with chromosome 22q11.2 deletion syndrome is associated with neurocognitive deficits and genetic polymorphisms. European Journal of Human Genetics, 2012, 20, 1051-1057.	1.4	17
49	Rare deleterious de novo missense variants in Rnf2/Ring2 are associated with a neurodevelopmental disorder with unique clinical features. Human Molecular Genetics, 2021, 30, 1283-1292.	1.4	17
50	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	0.6	16
51	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	16
52	De novo variants in <scp>TCF7L2</scp> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	0.7	13
53	A normative chart for cognitive development in a genetically selected population. Neuropsychopharmacology, 2022, 47, 1379-1386.	2.8	12
54	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11

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55	A pathogenic variant in the <scp>SETBP1</scp> hotspot results in a formeâ€fruste <scp>Schinzelâ€“Giedion</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1947-1951.	0.7	11
56	Communication of Psychiatric Risk in 22q11.2 Deletion Syndrome: A Pilot Project. Journal of Genetic Counseling, 2016, 25, 6-17.	0.9	9
57	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	1.4	8
58	Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity. American Journal of Human Genetics, 2022, 109, 518-532.	2.6	8
59	The genome empowerment scale: An assessment of parental empowerment in families with undiagnosed disease. Clinical Genetics, 2019, 96, 521-531.	1.0	7
60	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	1.4	7
61	Biâ€allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. Human Mutation, 2021, 42, 745-761.	1.1	7
62	Frontal Hypoactivation During a Working Memory Task in Children With 22q11 Deletion Syndrome. Journal of Child Neurology, 2017, 32, 94-99.	0.7	6
63	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	2.6	6
64	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
65	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	1.1	5
66	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	1.1	4
67	Epileptic encephalopathy with features of rapidâ€onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with ATP1A3 mutation. Epileptic Disorders, 2020, 22, 103-109.	0.7	4
68	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. Journal of Genetic Counseling, 2022, 31, 59-70.	0.9	3
69	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	3.7	3
70	Epilepsy in trisomy 7 mosaicism: A case report and literature review. Journal of Pediatric Neurology, 2015, 09, 063-068.	0.0	2
71	Completing the puzzle: The search for pieces in the understanding of psychosis risk in 22q11.2 deletion syndrome. Schizophrenia Research, 2017, 188, 33-34.	1.1	1
72	Hypogyrification and its association with cognitive impairment in children with 22q11.2 deletion Syndrome: A preliminary report. Psychiatry Research - Neuroimaging, 2019, 285, 47-50.	0.9	0