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	A normative chart for cognitive development in a genetically selected population. Neuropsychopharmacology, 2022, 47, 1379-1386.	2.8	12
2	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
3	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
4	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
5	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	1.1	5
6	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
7	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	3.7	3
8	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	1.1	4
9	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
10	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
11	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
12	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
13	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
14	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
15	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
16	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
17	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
18	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	1.1	16

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19	Genetic contributors to risk of schizophrenia in the presence of a 22q11.2 deletion. Molecular Psychiatry, 2021, 26, 4496-4510.	4.1	87
20	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
21	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
22	D-DEMÃ˜, a distinct phenotype caused by ATP1A3 mutations. Neurology: Genetics, 2020, 6, e466.	0.9	18
23	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
24	Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genetics in Medicine, 2020, 22, 1269-1275.	1.1	30
25	Phenotypic expansion of KMT2Dâ€related disorder: Beyond Kabuki syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1053-1065.	0.7	23
26	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
27	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
28	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
29	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
30	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
31	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
32	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
33	The genome empowerment scale: An assessment of parental empowerment in families with undiagnosed disease. Clinical Genetics, 2019, 96, 521-531.	1.0	7
34	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19
35	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
36	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

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37	Atypical chromosome 22q11.2 deletions are complex rearrangements and have different mechanistic origins. Human Molecular Genetics, 2019, 28, 3724-3733.	1.4	7
38	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
39	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
40	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
41	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
42	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
43	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	13.9	261
44	Characteristics of undiagnosed diseases network applicants: implications for referring providers. BMC Health Services Research, 2018, 18, 652.	0.9	23
45	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
46	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
47	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	1.4	54
48	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
49	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
50	Subthreshold Psychosis in 22q11.2 Deletion Syndrome: Multisite Naturalistic Study. Schizophrenia Bulletin, 2017, 43, 1079-1089.	2.3	47
51	The importance of dynamic re-analysis in diagnostic whole exome sequencing. Journal of Medical Genetics, 2017, 54, 155-156.	1.5	38
52	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
53	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
54	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

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55	Frontal Hypoactivation During a Working Memory Task in Children With 22q11 Deletion Syndrome. Journal of Child Neurology, 2017, 32, 94-99.	0.7	6
56	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	0.7	34
57	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
58	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
59	Communication of Psychiatric Risk in 22q11.2 Deletion Syndrome: A Pilot Project. Journal of Genetic Counseling, 2016, 25, 6-17.	0.9	9
60	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
61	Epilepsy in trisomy 7 mosaicism: A case report and literature review. Journal of Pediatric Neurology, 2015, 09, 063-068.	0.0	2
62	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
63	Quinidine in the treatment of <scp>KCNT</scp>1â€positive epilepsies. Annals of Neurology, 2015, 78, 995-999.	2.8	184
64	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
65	Whole-exome sequencing in undiagnosed genetic diseases: interpreting 119 trios. Genetics in Medicine, 2015, 17, 774-781.	1.1	284
66	Cognitive Decline Preceding the Onset of Psychosis in Patients With 22q11.2 Deletion Syndrome. JAMA Psychiatry, 2015, 72, 377.	6.0	196
67	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
68	Clinical application of exome sequencing in undiagnosed genetic conditions. Journal of Medical Genetics, 2012, 49, 353-361.	1.5	377
69	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
70	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
71	Evidence of gray matter reduction and dysfunction in chromosome 22q11.2 deletion syndrome. Psychiatry Research - Neuroimaging, 2010, 181, 1-8.	0.9	39
72	COMT and anxiety and cognition in children with chromosome 22q11.2 deletion syndrome. Psychiatry Research, 2010, 178, 433-436.	1.7	20