Pankaj B Agrawal

List of Publications by Year
in descending order

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92	2,454	236925	243625
papers	citations	25 h-index	44 g-index

94	94	94	3921
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
2	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
3	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	6.4	24
4	Perspectives of United States neonatologists on genetic testing practices. Genetics in Medicine, 2022, 24, 1372-1377.	2.4	6
5	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. JAMA Neurology, 2022, 79, 405.	9.0	7
6	Dynamin-2 reduction rescues the skeletal myopathy of a SPEG-deficient mouse model. JCI Insight, 2022, 7, .	5.0	5
7	A neurodevelopmental disorder caused by a novel de novo SVA insertion in exon 13 of the SRCAP gene. European Journal of Human Genetics, 2022, 30, 1083-1087.	2.8	8
8	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7
9	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
10	Exome sequencing identifies novel missense and deletion variants in <scp>RTN4IP1</scp> associated with optic atrophy, global developmental delay, epilepsy, ataxia, and choreoathetosis. American Journal of Medical Genetics, Part A, 2021, 185, 203-207.	1.2	5
11	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	7.9	10
12	Underrepresentation of Phenotypic Variability of 16p13.11 Microduplication Syndrome Assessed With an Online Self-Phenotyping Tool (Phenotypr): Cohort Study. Journal of Medical Internet Research, 2021, 23, e21023.	4.3	4
13	microRNA-mRNA Profile of Skeletal Muscle Differentiation and Relevance to Congenital Myotonic Dystrophy. International Journal of Molecular Sciences, 2021, 22, 2692.	4.1	5
14	Discordant results between conventional newborn screening and genomic sequencing in the BabySeq Project. Genetics in Medicine, 2021, 23, 1372-1375.	2.4	47
15	Homozygous SPEG Mutation Is Associated With Isolated Dilated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2021, 14, e003310.	3.6	9
16	Genetic Study in a Cohort of Children With ROHHAD Syndrome. Journal of the Endocrine Society, 2021, 5, A503-A504.	0.2	2
17	Striated Preferentially Expressed Protein Kinase (SPEG) in Muscle Development, Function, and Disease. International Journal of Molecular Sciences, 2021, 22, 5732.	4.1	12
18	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	13

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19	A homozygous stop-gain variant in ARHGAP42 is associated with childhood interstitial lung disease, systemic hypertension, and immunological findings. PLoS Genetics, 2021, 17, e1009639.	3.5	4
20	A data-driven architecture using natural language processing to improve phenotyping efficiency and accelerate genetic diagnoses of rare disorders. Human Genetics and Genomics Advances, 2021, 2, 100035.	1.7	4
21	Psychosocial Effect of Newborn Genomic Sequencing on Families in the BabySeq Project. JAMA Pediatrics, 2021, 175, 1132.	6.2	35
22	Neuroimaging in Kabuki syndrome and another <scp>KMT2D</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2021, 185, 3770-3783.	1.2	7
23	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1450-1465.	6.2	16
24	Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809.	1.2	4
25	SPEG binds with desmin and its deficiency causes defects in triad and focal adhesion proteins. Human Molecular Genetics, 2021, 29, 3882-3891.	2.9	6
26	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	8.2	53
27	Reanalysis of Exome Data Identifies Novel SLC25A46 Variants Associated with Leigh Syndrome. Journal of Personalized Medicine, 2021, 11, 1277.	2.5	2
28	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	2.5	17
29	Biallelic mutation of FBXL7 suggests a novel form of Hennekam syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 189-194.	1.2	13
30	Prospective, phenotype-driven selection of critically ill neonates for rapid exome sequencing is associated with high diagnostic yield. Genetics in Medicine, 2020, 22, 736-744.	2.4	83
31	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
32	De novo variants in MPP5 cause global developmental delay and behavioral changes. Human Molecular Genetics, 2020, 29, 3388-3401.	2.9	5
33	Familial and genetic factors in laryngeal cleft: Have we learned anything?. International Journal of Pediatric Otorhinolaryngology, 2020, 138, 110283.	1.0	0
34	Childrenâ€™s rare disease cohorts: an integrative research and clinical genomics initiative. Npj Genomic Medicine, 2020, 5, 29.	3.8	38
35	Heterozygous Variants in KDM4B Lead to Global Developmental Delay and Neuroanatomical Defects. American Journal of Human Genetics, 2020, 107, 1170-1177.	6.2	13
36	OR33-07 ARNT2: A Potential Novel Candidate Gene for Monogenic Obesity in Humans. Journal of the Endocrine Society, 2020, 4, .	0.2	0

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37	Congenital Heart Defects Due to TAF1 Missense Variants. Circulation Genomic and Precision Medicine, 2020, 13, e002843.	3.6	8
38	Knockin mouse model of the human CFL2 p.A35T mutation results in a unique splicing defect and severe myopathy phenotype. Human Molecular Genetics, 2020, 29, 1996-2003.	2.9	5
39	Reconsidering Genetic Testing for Neonatal Polycystic Kidney Disease. Kidney International Reports, 2020, 5, 1316-1319.	0.8	3
40	A phenotypically severe, biochemically â€œsilentâ€•case of HIBCH deficiency in a newborn diagnosed by rapid whole exome sequencing and enzymatic testing. American Journal of Medical Genetics, Part A, 2020, 182, 780-784.	1.2	4
41	Genetic diagnosis in the fetus. Journal of Perinatology, 2020, 40, 997-1006.	2.0	10
42	Quantifying Downstream Healthcare Utilization in Studies of Genomic Testing. Value in Health, 2020, 23, 559-565.	0.3	6
43	Prenatal Diagnosis of a Ventral Abdominal Wall Defect. NeoReviews, 2020, 21, e286-e292.	0.8	0
44	Striated Preferentially Expressed Protein Kinase (SPEG)-Deficient Skeletal Muscles Display Fewer Satellite Cells with Reduced Proliferation and Delayed Differentiation. American Journal of Pathology, 2020, 190, 2453-2463.	3.8	4
45	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	8.2	40
46	Deciphering congenital anomalies for the next generation. Journal of Physical Education and Sports Management, 2020, 6, a005504.	1.2	5
47	Expanding the phenotypic spectrum associated with OPHN1 variants. European Journal of Medical Genetics, 2019, 62, 137-143.	1.3	8
48	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
49	The impact of the Orphan Drug Act on Food and Drug Administration-approved therapies for rare skin diseases and skin-related cancers. Journal of the American Academy of Dermatology, 2019, 81, 867-877.	1.2	5
50	Etiology and Outcome of non-immune Hydrops Fetalis in Southern China: report of 1004 cases. Scientific Reports, 2019, 9, 10726.	3.3	10
51	Genome Sequencing Identifies the Pathogenic Variant Missed by Prior Testing in an Infant with Marfan Syndrome. Journal of Pediatrics, 2019, 213, 235-240.	1.8	6
52	Infant mortality: the contribution of genetic disorders. Journal of Perinatology, 2019, 39, 1611-1619.	2.0	47
53	Phenotype characterisation of TBX4 mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. European Respiratory Journal, 2019, 54, 1801965.	6.7	77
54	A novel missense mutation in TFAP2B associated with Char syndrome and central diabetes insipidus. American Journal of Medical Genetics, Part A, 2019, 179, 1299-1303.	1.2	4

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55	Unique bioinformatic approach and comprehensive reanalysis improve diagnostic yield of clinical exomes. European Journal of Human Genetics, 2019, 27, 1398-1405.	2.8	60
56	FDA oversight of NSIGHT genomic research: the need for an integrated systems approach to regulation. Npj Genomic Medicine, 2019, 4, 32.	3.8	6
57	Parental interest in genomic sequencing of newborns: enrollment experience from the BabySeq Project. Genetics in Medicine, 2019, 21, 622-630.	2.4	61
58	Chromosomal microarray and whole exome sequencing identify genetic causes of congenital hypothyroidism with extra-thyroidal congenital malformations. Clinica Chimica Acta, 2019, 489, 103-108.	1.1	7
59	Interpretation of Genomic Sequencing Results in Healthy and Ill Newborns: Results from the BabySeq Project. American Journal of Human Genetics, 2019, 104, 76-93.	6.2	176
60	Novel SPEG mutations in congenital myopathies: Genotypeâ€“phenotype correlations. Muscle and Nerve, 2019, 59, 357-362.	2.2	17
61	Mammalian Hbs1L deficiency causes congenital anomalies and developmental delay associated with Pelota depletion and 80S monosome accumulation. PLoS Genetics, 2019, 15, e1007917.	3.5	15
62	SPEG-deficient skeletal muscles exhibit abnormal triad and defective calcium handling. Human Molecular Genetics, 2018, 27, 1608-1617.	2.9	22
63	Genetic disorders and mortality in infancy and early childhood: delayed diagnoses and missed opportunities. Genetics in Medicine, 2018, 20, 1396-1404.	2.4	58
64	Threeâ€generation family with novel contiguous gene deletion on chromosome 2p22 associated with thoracic aortic aneurysm syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 560-569.	1.2	14
65	Characterization of a novel variant in siblings with Asparagine Synthetase Deficiency. Molecular Genetics and Metabolism, 2018, 123, 317-325.	1.1	23
66	Monogenic Hashimoto thyroiditis associated with a variant in the thyroglobulin (TG) gene. Journal of Autoimmunity, 2018, 86, 116-119.	6.5	13
67	De novo variant of TRRAP in a patient with very early onset psychosis in the context of non-verbal learning disability and obsessive-compulsive disorder: a case report. BMC Medical Genetics, 2018, 19, 197.	2.1	7
68	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
69	De novo variant in KIF26B is associated with pontocerebellar hypoplasia with infantile spinal muscular atrophy. American Journal of Medical Genetics, Part A, 2018, 176, 2623-2629.	1.2	19
70	De novo ATP1A3 and compound heterozygous NLRP3 mutations in a child with autism spectrum disorder, episodic fatigue and somnolence, and muckle-wells syndrome. Molecular Genetics and Metabolism Reports, 2018, 16, 23-29.	1.1	12
71	Pressure Overload in Mice With Haploinsufficiency of Striated Preferentially Expressed Gene Leads to Decompensated Heart Failure. Frontiers in Physiology, 2018, 9, 863.	2.8	7
72	The BabySeq project: implementing genomic sequencing in newborns. BMC Pediatrics, 2018, 18, 225.	1.7	115

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73	Novel founder intronic variant in SLC39A14 in two families causing Manganism and potential treatment strategies. Molecular Genetics and Metabolism, 2018, 124, 161-167.	1.1	36
74	Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report. Journal of Physical Education and Sports Management, 2018, 4, a002873.	1.2	7
75	Neonatal-Onset Chronic Diarrhea Caused by Homozygous Nonsense WNT2B Mutations. American Journal of Human Genetics, 2018, 103, 131-137.	6.2	16
76	A curated gene list for reporting results of newborn genomic sequencing. Genetics in Medicine, 2017, 19, 809-818.	2.4	79
77	Beta-Ketothiolase Deficiency Presenting with Metabolic Stroke After a Normal Newborn Screen in Two Individuals. JIMD Reports, 2017, 39, 45-54.	1.5	10
78	Heterozygous De Novo UBTF Gain-of-Function Variant Is Associated with Neurodegeneration in Childhood. American Journal of Human Genetics, 2017, 101, 267-273.	6.2	41
79	Novel mutation in CNTNAP1 results in congenital hypomyelinating neuropathy. Muscle and Nerve, 2017, 55, 761-765.	2.2	15
80	Exome sequencing results in successful diagnosis and treatment of a severe congenital anemia. Journal of Physical Education and Sports Management, 2016, 2, a000885.	1.2	10
81	Hyperammonemia as a Presenting Feature in Two Siblings with FBXL4 Variants. JIMD Reports, 2016, 35, 7-15.	1.5	9
82	A novel de novo mutation inATP1A3and childhood-onset schizophrenia. Journal of Physical Education and Sports Management, 2016, 2, a001008.	1.2	46
83	Clinical heterogeneity associated with KCNA1 mutations include cataplexy and nonataxic presentations. Neurogenetics, 2016, 17, 11-16.	1.4	26
84	Skeletal Muscle MicroRNA and Messenger RNA Profiling in Cofilin-2 Deficient Mice Reveals Cell Cycle Dysregulation Hindering Muscle Regeneration. PLoS ONE, 2015, 10, e0123829.	2.5	9
85	Expanding the Phenotype Associated With theNEFLMutation. JAMA Neurology, 2014, 71, 1413.	9.0	30
86	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. American Journal of Human Genetics, 2014, 95, 218-226.	6.2	143
87	Stimulating erythropoiesis in neonates. American Journal of Hematology, 2013, 88, 930-931.	4.1	1
88	Whole genome sequencing identifies <scp>SCN2A</scp> mutation in monozygotic twins with <scp>O</scp>htahara syndrome and unique neuropathologic findings. Epilepsia, 2013, 54, e81-5.	5.1	49
89	Rare complete loss of function provides insight into a pleiotropic genome-wide association study locus. Blood, 2013, 122, 3845-3847.	1.4	25
90	Normal myofibrillar development followed by progressive sarcomeric disruption with actin accumulations in a mouse Cfl2 knockout demonstrates requirement of cofilin-2 for muscle maintenance. Human Molecular Genetics, 2012, 21, 2341-2356.	2.9	80

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91	Nemaline Myopathy with Minicores Caused by Mutation of the CFL2 Gene Encoding the Skeletal Muscle Actinâ€“Binding Protein, Cofilin-2. American Journal of Human Genetics, 2007, 80, 162-167.	6.2	213
92	Heterogeneity of nemaline myopathy cases with skeletal muscle Î±â€actin gene mutations. Annals of Neurology, 2004, 56, 86-96.	5.3	135