Jamal Nasir

List of Publications by Year
in descending order

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45 papers	4,014 citations	257450 24 h-index	265206 42 g-index
46 all docs	46 docs citations	46 times ranked	4907 citing authors

#	Article	IF	Citations
1	SARS-CoV-2 Susceptibility and ACE2 Gene Variations Within Diverse Ethnic Backgrounds. Frontiers in Genetics, 2022, 13, 888025.	2.3	14
2	Mutations in MINAR2 encoding membrane integral NOTCH2-associated receptor 2 cause deafness in humans and mice. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	3
3	Whole exome sequencing reveals a homozygous SGCB variant in a Pakhtun family with limb girdle muscular dystrophy (LGMDR4) phenotype. Gene Reports, 2021, 22, 101014.	0.8	0
4	The Data Use Ontology to streamline responsible access to human biomedical datasets. Cell Genomics, 2021, 1, 100028.	6.5	31
5	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	6.5	94
6	Distinct proteomic profiles in monozygotic twins discordant for ischaemic stroke. Molecular and Cellular Biochemistry, 2019, 456, 157-165.	3.1	7
7	Overexpression of the dopamine receptorâ€interacting protein Alix/AIP1 modulatesNMDAreceptorâ€triggered cell death. FEBS Letters, 2019, 593, 1381-1391.	2.8	1
8	De novo single-nucleotide and copy number variation in discordant monozygotic twins reveals disease-related genes. European Journal of Human Genetics, 2019, 27, 1121-1133.	2.8	37
9	A mutation in the major autophagy gene, WIPI2, associated with global developmental abnormalities. Brain, 2019, 142, 1242-1254.	7.6	28
10	A missense mutation in TRAPPC6A leads to build-up of the protein, in patients with a neurodevelopmental syndrome and dysmorphic features. Scientific Reports, 2018, 8, 2053.	3.3	30
11	Case-based interprofessional learning for undergraduate healthcare professionals in the clinical setting. Journal of Interprofessional Care, 2017, 31, 125-128.	1.7	28
12	The alkylglycerol monooxygenase (AGMO) gene previously involved in autism also causes a novel syndromic form of primary microcephaly in a consanguineous Saudi family. Journal of the Neurological Sciences, 2016, 363, 240-244.	0.6	18
13	Identical non-identical twins and non-identical identical twins. BMJ, The, 2015, , h6589.	6.0	1
14	Exome analysis identified a novel missense mutation in the CLPP gene in a consanguineous Saudi family expanding the clinical spectrum of Perrault Syndrome type-3. Journal of the Neurological Sciences, 2015, 353, 149-154.	0.6	37
15	Elevated Î³-Glutamyltransferase and Erythrocyte Sedimentation Rate in Ischemic Stroke in Discordant Monozygotic Twin Study. International Journal of Stroke, 2015, 10, E32-E33.	5.9	1
16	Truncating mutation in intracellular phospholipase A1 gene (DDHD2) in hereditary spastic paraplegia with intellectual disability (SPG54). BMC Research Notes, 2015, 8, 271.	1.4	17
17	Clinically Significant Missense Variants in Human <scp>GALNT</scp>3, <scp>GALNT</scp>8, <scp>GALNT</scp>12, and <scp>GALNT</scp>13 Genes: Intriguing In Silico Findings. Journal of Cellular Biochemistry, 2014, 115, 313-327.	2.6	15
18	ZizB, a novel RacGEF regulates development, cell motility and cytokinesis in Dictyostelium.. Journal of Cell Science, 2012, 125, 2457-65.	2.0	15

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19	The Rac GEF ZizB regulates development, cell motility and cytokinesis in Dictyostelium. Development (Cambridge), 2012, 139, e1-e1.	2.5	0
20	CMIP and ATP2C2 Modulate Phonological Short-Term Memory in Language Impairment. American Journal of Human Genetics, 2009, 85, 264-272.	6.2	173
21	Abnormal vibration-induced illusion of movement in idiopathic focal dystonia: An endophenotypic marker?. Movement Disorders, 2008, 23, 373-377.	3.9	45
22	ALG-2 interacting protein AIP1: a novel link between D1 and D3 signalling. European Journal of Neuroscience, 2008, 27, 1626-1633.	2.6	19
23	Genetics of speech sounds great. Clinical Genetics, 2008, 54, 117-118.	2.0	0
24	Severe receptive language disorder in childhood--familial aspects and long-term outcomes: results from a Scottish study. Archives of Disease in Childhood, 2007, 92, 614-619.	1.9	43
25	Interaction between chromatin proteins MECP2 and ATRX is disrupted by mutations that cause inherited mental retardation. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 2709-2714.	7.1	231
26	Unbalanced whole arm translocation resulting in loss of 18p in dystonia. Movement Disorders, 2006, 21, 859-863.	3.9	22
27	Targeted disruption of Huntingtin-associated protein-1 (Hap1) results in postnatal death due to depressed feeding behavior. Human Molecular Genetics, 2002, 11, 945-959.	2.9	73
28	Cloning and Characterization of Three Novel Genes, ALS2CR1, ALS2CR2, and ALS2CR3, in the Juvenile Amyotrophic Lateral Sclerosis (ALS2) Critical Region at Chromosome 2q33â€“q34: Candidate Genes for ALS2. Genomics, 2001, 71, 200-213.	2.9	46
29	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	21.4	635
30	Life Without Huntingtin. Normal Differentiation into FunctionalNeurons. Journal of Neurochemistry, 1999, 72, 1009-1018.	3.9	37
31	Genomic organization of the human caspase-9 gene on Chromosome 1p36.1-p36.3. Mammalian Genome, 1999, 10, 757-760.	2.2	14
32	Neuronal degeneration in the basal ganglia and loss of pallido-subthalamic synapses in mice with targeted disruption of the Huntington's disease gene. Brain Research, 1999, 818, 468-479.	2.2	77
33	A YAC Mouse Model for Huntingtonâ€™s Disease with Full-Length Mutant Huntingtin, Cytoplasmic Toxicity, and Selective Striatal Neurodegeneration. Neuron, 1999, 23, 181-192.	8.1	789
34	A Yeast Artificial Chromosome-Based Physical Map of the Juvenile Amyotrophic Lateral Sclerosis (ALS2) Critical Region on Human Chromosome 2q33â€“q34. Genomics, 1999, 55, 106-112.	2.9	18
35	Cell death attenuation by `Usurpin', a mammalian DED-caspase homologue that precludes caspase-8 recruitment and activation by the CD-95 (Fas, APO-1) receptor complex. Cell Death and Differentiation, 1998, 5, 271-288.	11.2	293
36	Interleukin-1Î±-converting enzyme (ICE) and related cell death genes ICErel-II and ICErel-III map to the same PAC clone at band 11q22.2-22.3. Mammalian Genome, 1997, 8, 611-613.	2.2	11

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37	Huntington disease: new insights into the relationship between CAG expansion and disease. Human Molecular Genetics, 1996, 5, 1431-1435.	2.9	60
38	Structural analysis of the 5â€² region of mouse and human huntington disease genes reveals conservation of putative promoter region and di- and trinucleotide polymorphisms. Genomics, 1995, 25, 707-715.	2.9	41
39	Genomic organization of the human Î±-adducin gene and its alternately spliced isoforms. Genomics, 1995, 25, 93-99.	2.9	37
40	Targeted disruption of the Huntington's disease gene results in embryonic lethality and behavioral and morphological changes in heterozygotes. Cell, 1995, 81, 811-823.	28.9	758
41	The Murine Homologues of the Huntington Disease Gene (Hdh) and the Î±-Adducin Gene (Add1) Map to Mouse Chromosome 5 within a Region of Conserved Synteny with Human Chromosome 4p16.3. Genomics, 1994, 22, 198-201.	2.9	21
42	Murine Î±-l-Iduronidase: cDNA Isolation and Expression. Genomics, 1994, 24, 311-316.	2.9	30
43	Identification of an Alu retrotransposition event in close proximity to a strong candidate gene for Huntington's disease. Nature, 1993, 362, 370-373.	27.8	50
44	Differential 3â€² polyadenylation of the Huntington disease gene results in two mRNA species with variable tissue expression. Human Molecular Genetics, 1993, 2, 1541-1545.	2.9	94
45	Co-amplification of L1 line elements with localised low copy repeats in Giemsa dark bands: implications for genome organisation. Nucleic Acids Research, 1991, 19, 3255-3260.	14.5	19