

Jamal Nasir

List of Publications by Year in descending order

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#	ARTICLE	IF	CITATIONS
1	SARS-CoV-2 Susceptibility and ACE2 Gene Variations Within Diverse Ethnic Backgrounds. <i>Frontiers in Genetics</i> , 2022, 13, 888025.	1.1	14
2	Mutations in <i>MINAR2</i> encoding membrane integral NOTCH2-associated receptor 2 cause deafness in humans and mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	3
3	Whole exome sequencing reveals a homozygous <i>SCGB</i> variant in a Pakhtun family with limb girdle muscular dystrophy (<i>LGMDR4</i>) phenotype. <i>Gene Reports</i> , 2021, 22, 101014.	0.4	0
4	The Data Use Ontology to streamline responsible access to human biomedical datasets. <i>Cell Genomics</i> , 2021, 1, 100028.	3.0	31
5	GA4GH: International policies and standards for data sharing across genomic research and healthcare. <i>Cell Genomics</i> , 2021, 1, 100029.	3.0	94
6	Distinct proteomic profiles in monozygotic twins discordant for ischaemic stroke. <i>Molecular and Cellular Biochemistry</i> , 2019, 456, 157-165.	1.4	7
7	Overexpression of the dopamine receptor-interacting protein Alix/AIP1 modulates NMDA receptor-triggered cell death. <i>FEBS Letters</i> , 2019, 593, 1381-1391.	1.3	1
8	De novo single-nucleotide and copy number variation in discordant monozygotic twins reveals disease-related genes. <i>European Journal of Human Genetics</i> , 2019, 27, 1121-1133.	1.4	37
9	A mutation in the major autophagy gene, <i>WIPI2</i> , associated with global developmental abnormalities. <i>Brain</i> , 2019, 142, 1242-1254.	3.7	28
10	A missense mutation in <i>TRAPPC6A</i> leads to build-up of the protein, in patients with a neurodevelopmental syndrome and dysmorphic features. <i>Scientific Reports</i> , 2018, 8, 2053.	1.6	30
11	Case-based interprofessional learning for undergraduate healthcare professionals in the clinical setting. <i>Journal of Interprofessional Care</i> , 2017, 31, 125-128.	0.8	28
12	The alkylglycerol monooxygenase (<i>AGMO</i>) gene previously involved in autism also causes a novel syndromic form of primary microcephaly in a consanguineous Saudi family. <i>Journal of the Neurological Sciences</i> , 2016, 363, 240-244.	0.3	18
13	Identical non-identical twins and non-identical identical twins. <i>BMJ, The</i> , 2015, , h6589.	3.0	1
14	Exome analysis identified a novel missense mutation in the <i>CLPP</i> gene in a consanguineous Saudi family expanding the clinical spectrum of Perrault Syndrome type-3. <i>Journal of the Neurological Sciences</i> , 2015, 353, 149-154.	0.3	37
15	Elevated $\hat{3}$ -Glutamyltransferase and Erythrocyte Sedimentation Rate in Ischemic Stroke in Discordant Monozygotic Twin Study. <i>International Journal of Stroke</i> , 2015, 10, E32-E33.	2.9	1
16	Truncating mutation in intracellular phospholipase A1 gene (<i>DDHD2</i>) in hereditary spastic paraplegia with intellectual disability (<i>SPG54</i>). <i>BMC Research Notes</i> , 2015, 8, 271.	0.6	17
17	Clinically Significant Missense Variants in Human <i>GALNT3</i> , <i>GALNT8</i> , <i>GALNT12</i> , and <i>GALNT13</i> Genes: Intriguing In Silico Findings. <i>Journal of Cellular Biochemistry</i> , 2014, 115, 313-327.	1.2	15
18	ZizB, a novel RacGEF regulates development, cell motility and cytokinesis in <i>Dictyostelium</i> .. <i>Journal of Cell Science</i> , 2012, 125, 2457-65.	1.2	15

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19	The Rac GEF ZizB regulates development, cell motility and cytokinesis in Dictyostelium. <i>Development</i> (Cambridge), 2012, 139, e1-e1.	1.2	0
20	CMIP and ATP2C2 Modulate Phonological Short-Term Memory in Language Impairment. <i>American Journal of Human Genetics</i> , 2009, 85, 264-272.	2.6	173
21	Abnormal vibration-induced illusion of movement in idiopathic focal dystonia: An endophenotypic marker?. <i>Movement Disorders</i> , 2008, 23, 373-377.	2.2	45
22	ALG-2 interacting protein AIP1: a novel link between D1 and D3 signalling. <i>European Journal of Neuroscience</i> , 2008, 27, 1626-1633.	1.2	19
23	Genetics of speech sounds great. <i>Clinical Genetics</i> , 2008, 54, 117-118.	1.0	0
24	Severe receptive language disorder in childhood—familial aspects and long-term outcomes: results from a Scottish study. <i>Archives of Disease in Childhood</i> , 2007, 92, 614-619.	1.0	43
25	Interaction between chromatin proteins MECP2 and ATRX is disrupted by mutations that cause inherited mental retardation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 2709-2714.	3.3	231
26	Unbalanced whole arm translocation resulting in loss of 18p in dystonia. <i>Movement Disorders</i> , 2006, 21, 859-863.	2.2	22
27	Targeted disruption of Huntingtin-associated protein-1 (Hap1) results in postnatal death due to depressed feeding behavior. <i>Human Molecular Genetics</i> , 2002, 11, 945-959.	1.4	73
28	Cloning and Characterization of Three Novel Genes, ALS2CR1, ALS2CR2, and ALS2CR3, in the Juvenile Amyotrophic Lateral Sclerosis (ALS2) Critical Region at Chromosome 2q33-q34: Candidate Genes for ALS2. <i>Genomics</i> , 2001, 71, 200-213.	1.3	46
29	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. <i>Nature Genetics</i> , 2001, 29, 166-173.	9.4	635
30	Life Without Huntingtin. Normal Differentiation into Functional Neurons. <i>Journal of Neurochemistry</i> , 1999, 72, 1009-1018.	2.1	37
31	Genomic organization of the human caspase-9 gene on Chromosome 1p36.1-p36.3. <i>Mammalian Genome</i> , 1999, 10, 757-760.	1.0	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. <i>Brain Research</i> , 1999, 818, 468-479.	1.1	77
33	A YAC Mouse Model for Huntington's Disease with Full-Length Mutant Huntingtin, Cytoplasmic Toxicity, and Selective Striatal Neurodegeneration. <i>Neuron</i> , 1999, 23, 181-192.	3.8	789
34	A Yeast Artificial Chromosome-Based Physical Map of the Juvenile Amyotrophic Lateral Sclerosis (ALS2) Critical Region on Human Chromosome 2q33-q34. <i>Genomics</i> , 1999, 55, 106-112.	1.3	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. <i>Cell Death and Differentiation</i> , 1998, 5, 271-288.	5.0	293
36	Interleukin-1-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. <i>Mammalian Genome</i> , 1997, 8, 611-613.	1.0	11

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37	Huntington disease: new insights into the relationship between CAG expansion and disease. <i>Human Molecular Genetics</i> , 1996, 5, 1431-1435.	1.4	60
38	Structural analysis of the 5' region of mouse and human huntington disease genes reveals conservation of putative promoter region and di- and trinucleotide polymorphisms. <i>Genomics</i> , 1995, 25, 707-715.	1.3	41
39	Genomic organization of the human β -adducin gene and its alternately spliced isoforms. <i>Genomics</i> , 1995, 25, 93-99.	1.3	37
40	Targeted disruption of the Huntington's disease gene results in embryonic lethality and behavioral and morphological changes in heterozygotes. <i>Cell</i> , 1995, 81, 811-823.	13.5	758
41	The Murine Homologues of the Huntington Disease Gene (Hdh) and the β -Adducin Gene (Add1) Map to Mouse Chromosome 5 within a Region of Conserved Synteny with Human Chromosome 4p16.3. <i>Genomics</i> , 1994, 22, 198-201.	1.3	21
42	Murine β -Iduronidase: cDNA Isolation and Expression. <i>Genomics</i> , 1994, 24, 311-316.	1.3	30
43	Identification of an Alu retrotransposition event in close proximity to a strong candidate gene for Huntington's disease. <i>Nature</i> , 1993, 362, 370-373.	13.7	50
44	Differential 3' polyadenylation of the Huntington disease gene results in two mRNA species with variable tissue expression. <i>Human Molecular Genetics</i> , 1993, 2, 1541-1545.	1.4	94
45	Co-amplification of L1 line elements with localised low copy repeats in Giemsa dark bands: implications for genome organisation. <i>Nucleic Acids Research</i> , 1991, 19, 3255-3260.	6.5	19