S Rangasamy

List of Publications by Year
in descending order

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49	2,409	331259	253896
papers	citations	21 h-index	43 g-index

55	55	55	4427
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Progressive cerebellar atrophy caused by heterozygous TECPR2 mutations. Molecular Genetics & Genomic Medicine, 2022, 10, e1857.	0.6	1
2	eP329: Genome sequencing uncovers molecular cause in a case with epileptic encephalopathy. Genetics in Medicine, 2022, 24, S206.	1.1	0
3	Okur-Chung neurodevelopmental syndrome-linked CK2Î± variants have reduced kinase activity. Human Genetics, 2021, 140, 1077-1096.	1.8	11
4	Improved methods for RNAseq-based alternative splicing analysis. Scientific Reports, 2021, 11, 10740.	1.6	22
5	Congenital myasthenic syndrome caused by a frameshift insertion mutation in GFPT1. Neurology: Genetics, 2020, 6, e468.	0.9	8
6	Transcriptomics analysis of pericytes from retinas of diabetic animals reveals novel genes and molecular pathways relevant to blood-retinal barrier alterations in diabetic retinopathy. Experimental Eye Research, 2020, 195, 108043.	1.2	17
7	Genes Implicated in Rare Congenital Inner Ear and Cochleovestibular Nerve Malformations. Ear and Hearing, 2020, 41, 983-989.	1.0	6
8	Do Genomic Factors Play a Role in Diabetic Retinopathy?. Journal of Clinical Medicine, 2020, 9, 216.	1.0	28
9	Genotypes and Phenotypes: A Search for Influential Genes in Diabetic Retinopathy. International Journal of Molecular Sciences, 2020, 21, 2712.	1.8	20
10	Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525.	2.6	50
11	Two additional males with Xâ€linked, syndromic mental retardation carry de novo mutations in HNRNPH2. Clinical Genetics, 2019, 96, 183-185.	1.0	16
12	A de novo SIX1 variant in a patient with a rare nonsyndromic cochleovestibular nerve abnormality, cochlear hypoplasia, and bilateral sensorineural hearing loss. Molecular Genetics & Genomic Medicine, 2019, 7, e995.	0.6	6
13	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. Nature Communications, 2019, 10, 708.	5.8	40
14	Compound heterozygous mutations in SNAP29 is associated with Pelizaeus-Merzbacher-like disorder (PMLD). Human Genetics, 2019, 138, 1409-1417.	1.8	14
15	Neonatal epileptic encephalopathy caused by de novo GNAO1 mutation misdiagnosed as atypical Rett syndrome: Cautions in interpretation of genomic test results. Seminars in Pediatric Neurology, 2018, 26, 28-32.	1.0	10
16	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.	0.7	19
17	A novel FBXO28 frameshift mutation in a child with developmental delay, dysmorphic features, and intractable epilepsy: A second gene that may contribute to the 1q41â€q42 deletion phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1549-1558.	0.7	9
18	A de novo splice site mutation in CASK causes FG syndromeâ€4 and congenital nystagmus. American Journal of Medical Genetics, Part A, 2017, 173, 611-617.	0.7	11

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19	Association of increased levels of MCP-1 and cathepsin-D in young onset type 2 diabetes patients (T2DM-Y) with severity of diabetic retinopathy. Journal of Diabetes and Its Complications, 2017, 31, 804-809.	1.2	26
20	Exploring genome-wide DNA methylation patterns in Aicardi syndrome. Epigenomics, 2017, 9, 1373-1386.	1.0	8
21	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
22	Phenotypic Variability and mTOR Pathway Gene Aberrations in Familial Tuberous Sclerosis. Journal of Pediatric Neurology, 2017, 15, 316-324.	0.0	0
23	Cellular and Animal Models of Neurologic Disease. , 2017, , 114-122.		0
24	Case Report: Novel mutations in TBC1D24 are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. F1000Research, 2017, 6, 553.	0.8	24
25	Dystonia in ATP2B3â€associated Xâ€linked spinocerebellar ataxia. Movement Disorders, 2016, 31, 1752-1753.	2.2	19
26	A de novo missense mutation in ZMYND11 is associated with global developmental delay, seizures, and hypotonia. Journal of Physical Education and Sports Management, 2016, 2, a000851.	0.5	16
27	Reduced neuronal size and mTOR pathway activity in the Mecp2 A140V Rett syndrome mouse model. F1000Research, 2016, 5, 2269.	0.8	38
28	Incontinentia pigmenti (Blochâ€“Sulzberger syndrome). Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2015, 132, 271-280.	1.0	17
29	A De Novo Mutation inTEAD1Causes Nonâ€“X-Linked Aicardi Syndrome. , 2015, 56, 3896.		22
30	Diabetic Macular Edema: Pathophysiology and Novel Therapeutic Targets. Ophthalmology, 2015, 122, 1375-1394.	2.5	396
31	New treatments for diabetic retinopathy. Diabetes, Obesity and Metabolism, 2015, 17, 219-230.	2.2	62
32	Chemokine Mediated Monocyte Trafficking into the Retina: Role of Inflammation in Alteration of the Blood-Retinal Barrier in Diabetic Retinopathy. PLoS ONE, 2014, 9, e108508.	1.1	186
33	Epigenetics, Autism Spectrum, and Neurodevelopmental Disorders. Neurotherapeutics, 2013, 10, 742-756.	2.1	100
34	The role of monocyte subsets in myocutaneous revascularization. Journal of Surgical Research, 2013, 183, 963-975.	0.8	17
35	Molecular Intricacies and the Role of ER Stress in Diabetes. Experimental Diabetes Research, 2012, 2012, 1-2.	3.8	10
36	Diabetic retinopathy and inflammation: Novel therapeutic targets. Middle East African Journal of Ophthalmology, 2012, 19, 52.	0.5	184

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37	A Potential Role for Angiopoietin 2 in the Regulation of the Bloodâ€“Retinal Barrier in Diabetic Retinopathy. , 2011, 52, 3784.		131
38	Pericyte-Derived Sphinogosine 1-Phosphate Induces the Expression of Adhesion Proteins and Modulates the Retinal Endothelial Cell Barrier. Arteriosclerosis, Thrombosis, and Vascular Biology, 2011, 31, e107-15.	1.1	49
39	Oxidative stress is independently associated with non-alcoholic fatty liver disease (NAFLD) in subjects with and without type 2 diabetes. Clinical Biochemistry, 2010, 43, 815-821.	0.8	96
40	Association of Leukocyte Count With Varying Degrees of Glucose Intolerance in Asian Indians: The Chennai Urban Rural Epidemiology Study (CURES-26). Metabolic Syndrome and Related Disorders, 2009, 7, 205-210.	0.5	19
41	Advanced glycation index and its association with severity of diabetic retinopathy in type 2 diabetic subjects. Journal of Diabetes and Its Complications, 2008, 22, 261-266.	1.2	20
42	A Common Nonsynonymous Single Nucleotide Polymorphism in the SLC30A8 Gene Determines ZnT8 Autoantibody Specificity in Type 1 Diabetes. Diabetes, 2008, 57, 2693-2697.	0.3	186
43	Glutamine fructose-6-phosphate amidotransferase (GFAT) gene expression and activity in patients with type 2 diabetes: Inter-relationships with hyperglycaemia and oxidative stress. Clinical Biochemistry, 2007, 40, 952-957.	0.8	55
44	Association of hypoglutathionemia with reduced Na+/K+ ATPase activity in type 2 diabetes and microangiopathy. Molecular and Cellular Biochemistry, 2006, 282, 169-176.	1.4	21
45	Increased glutathionylated hemoglobin (HbSSG) in type 2 diabetes subjects with microangiopathy. Clinical Biochemistry, 2005, 38, 892-899.	0.8	66
46	Is insulin signaling molecules misguided in diabetes for ubiquitinâ€“proteasome mediated degradation?. Molecular and Cellular Biochemistry, 2005, 275, 117-125.	1.4	33
47	A novel advanced glycation index and its association with diabetes and microangiopathy. Metabolism: Clinical and Experimental, 2005, 54, 1002-1007.	1.5	47
48	Curcumin-induced inhibition of cellular reactive oxygen species generation: Novel therapeutic implications. Journal of Biosciences, 2003, 28, 715-721.	0.5	198
49	Modeling of Pontocerebellar Hypoplasia Type 1B and Chemical Mimicry in Patient-Derived Neural Stem Cells. SSRN Electronic Journal, 0, , .	0.4	0