

Frederic Laumonnier

List of Publications by Year in descending order

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48
papers

3,748
citations

257450

24
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197818

49
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all docs

52
docs citations

52
times ranked

6833
citing authors

#	ARTICLE	IF	CITATIONS
1	Identification of Novel Gene Variants for Autism Spectrum Disorders in the Lebanese Population Using Whole-Exome Sequencing. <i>Genes</i> , 2022, 13, 186.	2.4	5
2	<i>SEMA6B</i> variants cause intellectual disability and alter dendritic spine density and axon guidance. <i>Human Molecular Genetics</i> , 2022, 31, 3325-3340.	2.9	5
3	Genes containing hexanucleotide repeats resembling C9ORF72 and expressed in the central nervous system are frequent in the human genome. <i>Neurobiology of Aging</i> , 2021, 97, 148.e1-148.e7.	3.1	1
4	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. <i>Genetics in Medicine</i> , 2021, 23, 352-362.	2.4	23
5	Effect of familial clustering in the genetic screening of 235 French ALS families. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 479-484.	1.9	7
6	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. <i>Human Genetics</i> , 2021, 140, 885-896.	3.8	10
7	Dysregulations of Expression of Genes of the Ubiquitin/SUMO Pathways in an In Vitro Model of Amyotrophic Lateral Sclerosis Combining Oxidative Stress and SOD1 Gene Mutation. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1796.	4.1	10
8	Novel missense mutations in PTCHD1 alter its plasma membrane subcellular localization and cause intellectual disability and autism spectrum disorder. <i>Human Mutation</i> , 2021, 42, 848-861.	2.5	8
9	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. <i>American Journal of Human Genetics</i> , 2021, 108, 951-961.	6.2	26
10	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 356-370.	6.2	171
11	A novel mutation in the cleavage site N291 of TDP-43 protein in a familial case of amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 463-466.	1.7	1
12	Identification of rare copy number variations reveals PJA2 , APCS , SYNPO , and TAC1 as novel candidate genes in Autism Spectrum Disorders. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e786.	1.2	12
13	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. <i>Human Mutation</i> , 2019, 40, 2021-2032.	2.5	42
14	LIMK2-1 is a Hominidae-Specific Isoform of LIMK2 Expressed in Central Nervous System and Associated with Intellectual Disability. <i>Neuroscience</i> , 2019, 399, 199-210.	2.3	6
15	Post hoc analysis of plasma amino acid profiles: towards a specific pattern in autism spectrum disorder and intellectual disability. <i>Annals of Clinical Biochemistry</i> , 2018, 55, 543-552.	1.6	18
16	A novel mutation in the transmembrane 6 domain of <i>GABBR2</i> leads to a Rett-like phenotype. <i>Annals of Neurology</i> , 2018, 83, 437-439.	5.3	19
17	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. <i>Molecular Psychiatry</i> , 2016, 21, 133-148.	7.9	243
18	GABA/Glutamate synaptic pathways targeted by integrative genomic and electrophysiological explorations distinguish autism from intellectual disability. <i>Molecular Psychiatry</i> , 2016, 21, 411-418.	7.9	31

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19	Disruption of TCA Cycle and Glutamate Metabolism Identified by Metabolomics in an In Vitro Model of Amyotrophic Lateral Sclerosis. <i>Molecular Neurobiology</i> , 2016, 53, 6910-6924.	4.0	37
20	Mutation screening of the ubiquitin ligase gene RNF135 in French patients with autism. <i>Psychiatric Genetics</i> , 2015, 25, 263-267.	1.1	18
21	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2015, 96, 386-396.	6.2	27
22	Phenotypic spectrum associated with <i>PTCHD1</i> deletions and truncating mutations includes intellectual disability and autism spectrum disorder. <i>Clinical Genetics</i> , 2015, 88, 224-233.	2.0	63
23	Advances in Cellular Models to Explore the Pathophysiology of Amyotrophic Lateral Sclerosis. <i>Molecular Neurobiology</i> , 2014, 49, 966-983.	4.0	19
24	Rescue of fragile X syndrome phenotypes in Fmr1KO mice by a BKCa channel opener molecule. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 124.	2.7	92
25	ZC4H2 Mutations Are Associated with Arthrogyriposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 681-695.	6.2	68
26	¹ H- ¹³ C NMR-based urine metabolic profiling in autism spectrum disorders. <i>Talanta</i> , 2013, 114, 95-102.	5.5	79
27	GC-MS-based urine metabolic profiling of autism spectrum disorders. <i>Analytical and Bioanalytical Chemistry</i> , 2013, 405, 5291-5300.	3.7	109
28	Xq27 FRAXA Locus is a Strong Candidate for Dyslexia: Evidence from a Genome-Wide Scan in French Families. <i>Behavior Genetics</i> , 2013, 43, 132-140.	2.1	8
29	LIMK2d, a truncated isoform of Lim kinase 2 regulates neurite growth in absence of the LIM kinase domain. <i>Biochemical and Biophysical Research Communications</i> , 2012, 420, 247-252.	2.1	5
30	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. <i>Molecular Psychiatry</i> , 2012, 17, 1103-1115.	7.9	97
31	FG syndrome: The FGS2 locus revisited. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1489-1492.	1.2	3
32	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. <i>BMC Medical Genetics</i> , 2011, 12, 17.	2.1	25
33	A novel mutation in the DLG3 gene encoding the synapse-associated protein 102 (SAP102) causes non-syndromic mental retardation. <i>Neurogenetics</i> , 2010, 11, 251-255.	1.4	49
34	Mutations of the UPF3B gene, which encodes a protein widely expressed in neurons, are associated with nonspecific mental retardation with or without autism. <i>Molecular Psychiatry</i> , 2010, 15, 767-776.	7.9	113
35	Could autism with mental retardation result from digenism and frequent de novo mutations?. <i>World Journal of Biological Psychiatry</i> , 2009, 10, 1030-1036.	2.6	5
36	Recurrent Rearrangements in Synaptic and Neurodevelopmental Genes and Shared Biologic Pathways in Schizophrenia, Autism, and Mental Retardation. <i>Archives of General Psychiatry</i> , 2009, 66, 947.	12.3	374

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37	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	3.8	24
38	Autism and Nonsyndromic Mental Retardation Associated with a De Novo Mutation in the NLGN4X Gene Promoter Causing an Increased Expression Level. Biological Psychiatry, 2009, 66, 906-910.	1.3	61
39	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	6.2	187
40	Drs. Laumonnier, Le Guennec, Roger, and Briault Reply. American Journal of Psychiatry, 2007, 164, 978-979.	7.2	1
41	The Role of Neuronal Complexes in Human X-Linked Brain Diseases. American Journal of Human Genetics, 2007, 80, 205-220.	6.2	100
42	Homozygous silencing of T-box transcription factor EOMES leads to microcephaly with polymicrogyria and corpus callosum agenesis. Nature Genetics, 2007, 39, 454-456.	21.4	181
43	Drs. Laumonnier, Le Guennec, Roger, and Briault Reply. American Journal of Psychiatry, 2007, 164, 978.	7.2	0
44	Association of a Functional Deficit of the BK _{Ca} Channel, a Synaptic Regulator of Neuronal Excitability, With Autism and Mental Retardation. American Journal of Psychiatry, 2006, 163, 1622-1629.	7.2	158
45	Association of a Functional Deficit of the BK _{Ca} Channel, a Synaptic Regulator of Neuronal Excitability, With Autism and Mental Retardation. American Journal of Psychiatry, 2006, 163, 1622.	7.2	46
46	Mutations in PHF8 are associated with X linked mental retardation and cleft lip/cleft palate. Journal of Medical Genetics, 2005, 42, 780-786.	3.2	194
47	X-Linked Mental Retardation and Autism Are Associated with a Mutation in the NLGN4 Gene, a Member of the Neuroligin Family. American Journal of Human Genetics, 2004, 74, 552-557.	6.2	686
48	Transcription Factor SOX3 Is Involved in X-Linked Mental Retardation with Growth Hormone Deficiency. American Journal of Human Genetics, 2002, 71, 1450-1455.	6.2	265