Frederic Laumonnier

List of Publications by Year
in descending order

48 papers	3,748 citations	257450 24 h-index	197818 49 g-index
52 all docs	52 docs citations	52 times ranked	6833 citing authors

#	Article	IF	Citations
1	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
2	Recurrent Rearrangements in Synaptic and Neurodevelopmental Genes and Shared Biologic Pathways in Schizophrenia, Autism, and Mental Retardation. Archives of General Psychiatry, 2009, 66, 947.	12.3	374
3	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
4	X-exome sequencing of 405 unresolved families identifies seven novel intellectual disability genes. Molecular Psychiatry, 2016, 21, 133-148.	7.9	243
5	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
6	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
7	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
8	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
9	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
10	Mutations of the UPF3B gene, which encodes a protein widely expressed in neurons, are associated with nonspecific mental retardation with or without autism. Molecular Psychiatry, 2010, 15, 767-776.	7.9	113
11	GC-MS-based urine metabolic profiling of autism spectrum disorders. Analytical and Bioanalytical Chemistry, 2013, 405, 5291-5300.	3.7	109
12	The Role of Neuronal Complexes in Human X-Linked Brain Diseases. American Journal of Human Genetics, 2007, 80, 205-220.	6.2	100
13	Transcriptome profiling of UPF3B/NMD-deficient lymphoblastoid cells from patients with various forms of intellectual disability. Molecular Psychiatry, 2012, 17, 1103-1115.	7.9	97
14	Rescue of fragile X syndrome phenotypes in Fmr1KO mice by a BKCa channel opener molecule. Orphanet Journal of Rare Diseases, 2014, 9, 124.	2.7	92
15	1Hâ€“13C NMR-based urine metabolic profiling in autism spectrum disorders. Talanta, 2013, 114, 95-102.	5.5	79
16	ZC4H2 Mutations Are Associated with Arthrogryposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. American Journal of Human Genetics, 2013, 92, 681-695.	6.2	68
17	Phenotypic spectrum associated with <scp>PTCHD1</scp> deletions and truncating mutations includes intellectual disability and autism spectrum disorder. Clinical Genetics, 2015, 88, 224-233.	2.0	63
18	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

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19	A novel mutation in the DLG3 gene encoding the synapse-associated protein 102 (SAP102) causes non-syndromic mental retardation. Neurogenetics, 2010, 11, 251-255.	1.4	49
20	Association of a Functional Deficit of the BK<char aid="99823229" id="sub">Ca</char> Channel, a Synaptic Regulator of Neuronal Excitability, With Autism and Mental Retardation. American Journal of Psychiatry, 2006, 163, 1622.	7.2	46
21	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. Human Mutation, 2019, 40, 2021-2032.	2.5	42
22	Disruption of TCA Cycle and Glutamate Metabolism Identified by Metabolomics in an In Vitro Model of Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2016, 53, 6910-6924.	4.0	37
23	GABA/Glutamate synaptic pathways targeted by integrative genomic and electrophysiological explorations distinguish autism from intellectual disability. Molecular Psychiatry, 2016, 21, 411-418.	7.9	31
24	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	6.2	27
25	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	6.2	26
26	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. BMC Medical Genetics, 2011, 12, 17.	2.1	25
27	Haploinsufficiency of the GPD2 gene in a patient with nonsyndromic mental retardation. Human Genetics, 2009, 124, 649-658.	3.8	24
28	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	2.4	23
29	Advances in Cellular Models to Explore the Pathophysiology of Amyotrophic Lateral Sclerosis. Molecular Neurobiology, 2014, 49, 966-983.	4.0	19
30	A novel mutation in the transmembrane 6 domain of <i>GABBR2</i> leads to a Rettâ€like phenotype. Annals of Neurology, 2018, 83, 437-439.	5.3	19
31	Mutation screening of the ubiquitin ligase gene RNF135 in French patients with autism. Psychiatric Genetics, 2015, 25, 263-267.	1.1	18
32	Post hoc analysis of plasma amino acid profiles: towards a specific pattern in autism spectrum disorder and intellectual disability. Annals of Clinical Biochemistry, 2018, 55, 543-552.	1.6	18
33	Identification of rare copy number variations reveals PJA2 , APCS , SYNPO , and TAC1 as novel candidate genes in Autism Spectrum Disorders. Molecular Genetics & Genomic Medicine, 2019, 7, e786.	1.2	12
34	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. Human Genetics, 2021, 140, 885-896.	3.8	10
35	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. International Journal of Molecular Sciences, 2021, 22, 1796.	4.1	10
36	Xq27 FRAXA Locus is a Strong Candidate for Dyslexia: Evidence from a Genome-Wide Scan in French Families. Behavior Genetics, 2013, 43, 132-140.	2.1	8

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37	Novel missense mutations in PTCHD1 alter its plasma membrane subcellular localization and cause intellectual disability and autism spectrum disorder. Human Mutation, 2021, 42, 848-861.	2.5	8
38	Effect of familial clustering in the genetic screening of 235 French ALS families. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 479-484.	1.9	7
39	LIMK2-1 is a Hominidae-Specific Isoform of LIMK2 Expressed in Central Nervous System and Associated with Intellectual Disability. Neuroscience, 2019, 399, 199-210.	2.3	6
40	Could autism with mental retardation result from digenism and frequent de novo mutations?. World Journal of Biological Psychiatry, 2009, 10, 1030-1036.	2.6	5
41	LIMK2d, a truncated isoform of Lim kinase 2 regulates neurite growth in absence of the LIM kinase domain. Biochemical and Biophysical Research Communications, 2012, 420, 247-252.	2.1	5
42	Identification of Novel Gene Variants for Autism Spectrum Disorders in the Lebanese Population Using Whole-Exome Sequencing. Genes, 2022, 13, 186.	2.4	5
43	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
44	FG syndrome: The FGS2 locus revisited. American Journal of Medical Genetics, Part A, 2012, 158A, 1489-1492.	1.2	3
45	Drs. Laumonnier, Le Guennec, Roger, and Briault Reply. American Journal of Psychiatry, 2007, 164, 978-979.	7.2	1
46	A novel mutation in the cleavage site N291 of TDP-43 protein in a familial case of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 463-466.	1.7	1
47	Genes containing hexanucleotide repeats resembling C9ORF72 and expressed in the central nervous system are frequent in the human genome. Neurobiology of Aging, 2021, 97, 148.e1-148.e7.	3.1	1
48	Drs. Laumonnier, Le Guennec, Roger, and Briault Reply. American Journal of Psychiatry, 2007, 164, 978.	7.2	0