Laurent Villard

List of Publications by Year
in descending order

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114	5,464	94269	95083
papers	citations	37	68
		h-index	g-index

225	225	225	9584
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in a putative global transcriptional regulator cause X-linked mental retardation with Î±-thalassemia (ATR-X syndrome). Cell, 1995, 80, 837-845.	13.5	583
2	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	9.4	431
3	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	3.7	426
4	Mecp2 Deficiency Disrupts Norepinephrine and Respiratory Systems in Mice. Journal of Neuroscience, 2005, 25, 11521-11530.	1.7	251
5	MECP2 mutations in males. Journal of Medical Genetics, 2007, 44, 417-423.	1.5	150
6	Treatment with desipramine improves breathing and survival in a mouse model for Rett syndrome. European Journal of Neuroscience, 2007, 25, 1915-1922.	1.2	126
7	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. Brain, 2010, 133, 3194-3209.	3.7	125
8	Epileptic and nonepileptic features in patients with early onset epileptic encephalopathy and STXBP1 mutations. Epilepsia, 2011, 52, 1828-1834.	2.6	116
9	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	2.6	112
10	A Locus for Bilateral Perisylvian Polymicrogyria Maps to Xq28. American Journal of Human Genetics, 2002, 70, 1003-1008.	2.6	111
11	Novel Compound Heterozygous Mutations inTBC1D24Cause Familial Malignant Migrating Partial Seizures of Infancy. Human Mutation, 2013, 34, 869-872.	1.1	110
12	XNP mutation in a large family with Juberg-Marsidi syndrome. Nature Genetics, 1996, 12, 359-360.	9.4	101
13	Craniofacial expression of human and murine TBX22 correlates with the cleft palate and ankyloglossia phenotype observed in CPX patients. Human Molecular Genetics, 2002, 11, 2793-2804.	1.4	87
14	Similar early characteristics but variable neurological outcome of patients with a de novo mutation of KCNQ2. Orphanet Journal of Rare Diseases, 2013, 8, 80.	1.2	82
15	A cluster of translocation breakpoints in 2q37 is associated with overexpression of NPPC in patients with a similar overgrowth phenotype. Human Mutation, 2007, 28, 1183-1188.	1.1	79
16	Brain magnetic resonance study of Mecp2 deletion effects on anatomy and metabolism. Biochemical and Biophysical Research Communications, 2006, 340, 776-783.	1.0	75
17	Modification of Mecp2 dosage alters axonal transport through the Huntingtin/Hap1 pathway. Neurobiology of Disease, 2012, 45, 786-795.	2.1	68
18	Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. Human Molecular Genetics, 2013, 22, 3306-3314.	1.4	62

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19	Clinical and molecular findings in 39 patients with KBG syndrome caused by deletion or mutation of ANKRD11. American Journal of Medical Genetics, Part A, 2016, 170, 2847-2859.	0.7	62
20	Determination of the Genomic Structure of the XNP/ATRX Gene Encoding a Potential Zinc Finger Helicase. Genomics, 1997, 43, 149-155.	1.3	60
21	A recurrent KCNQ2 pore mutation causing early onset epileptic encephalopathy has a moderate effect on M current but alters subcellular localization of Kv7 channels. Neurobiology of Disease, 2015, 80, 80-92.	2.1	59
22	Disruption of the ATP8A2 gene in a patient with a t(10;13) de novo balanced translocation and a severe neurological phenotype. European Journal of Human Genetics, 2010, 18, 1360-1363.	1.4	58
23	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
24	TCF4Deletions in Pitt-Hopkins Syndrome. Human Mutation, 2008, 29, E242-E251.	1.1	53
25	Mutations in BCAP31 Cause a Severe X-Linked Phenotype with Deafness, Dystonia, and Central Hypomyelination and Disorganize the Golgi Apparatus. American Journal of Human Genetics, 2013, 93, 579-586.	2.6	53
26	Metabolic Fingerprints of Altered Brain Growth, Osmoregulation and Neurotransmission in a Rett Syndrome Model. PLoS ONE, 2007, 2, e157.	1.1	50
27	AP1S2 is mutated in X-linked Dandyâ€“Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). European Journal of Human Genetics, 2014, 22, 363-368.	1.4	49
28	Tyrosine hydroxylase deficit in the chemoafferent and the sympathoadrenergic pathways of the Mecp2 deficient mouse. Neuroscience Letters, 2008, 447, 82-86.	1.0	48
29	Morphological and functional alterations in the substantia nigra pars compacta of the Mecp2-null mouse. Neurobiology of Disease, 2011, 41, 385-397.	2.1	48
30	GABA and Glutamate Pathways Are Spatially and Developmentally Affected in the Brain of Mecp2-Deficient Mice. PLoS ONE, 2014, 9, e92169.	1.1	48
31	The phenotypic spectrum of WWOX-related disorders: 20 additional cases of WOREE syndrome and review of the literature. Genetics in Medicine, 2019, 21, 1308-1318.	1.1	48
32	Carpenter-Waziri syndrome results from a mutation in XNP. , 1999, 85, 249-251.		47
33	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. Journal of Medical Genetics, 2010, 47, 155-161.	1.5	47
34	Epilepsy in Rett syndromeâ€”Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	2.6	47
35	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
36	The Incidence of Rett Syndrome in France. Pediatric Neurology, 2006, 34, 372-375.	1.0	44

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37	Epileptic patients with de novo <scp>STXBP</scp>1 mutations: Key clinical features based on 24 cases. Epilepsia, 2015, 56, 1931-1940.	2.6	44
38	Variable clinical expression in patients with mosaicism for KCNQ2 mutations. American Journal of Medical Genetics, Part A, 2015, 167, 2314-2318.	0.7	41
39	Exome sequencing in congenital ataxia identifies two new candidate genes and highlights a pathophysiological link between some congenital ataxias and early infantile epileptic encephalopathies. Genetics in Medicine, 2019, 21, 553-563.	1.1	41
40	Linkage of X-linked myopathy with excessive autophagy (XMEA) to Xq28. European Journal of Human Genetics, 2000, 8, 125-129.	1.4	39
41	Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768.	1.8	38
42	Identification of a mutation in theXNP/ATR-X gene in a family reported as Smith-Fineman-Myers syndrome. , 2000, 91, 83-85.		37
43	Deletion of YWHAE in a patient with periventricular heterotopias and pronounced corpus callosum hypoplasia. Journal of Medical Genetics, 2010, 47, 132-136.	1.5	37
44	Biogenic amines and their metabolites are differentially affected in the Mecp2-deficient mouse brain. BMC Neuroscience, 2011, 12, 47.	0.8	35
45	Evidence that homozygous PTPRD gene microdeletion causes trigonocephaly, hearing loss, and intellectual disability. Molecular Cytogenetics, 2015, 8, 39.	0.4	35
46	A codon-optimized Mecp2 transgene corrects breathing deficits and improves survival in a mouse model of Rett syndrome. Neurobiology of Disease, 2017, 99, 1-11.	2.1	35
47	Biogenic Amines in Rett Syndrome: The Usual Suspects. Behavior Genetics, 2010, 40, 59-75.	1.4	34
48	A C2055T Transition in Exon 8 of the ATP7A Gene Is Associated with Exon Skipping in an Occipital Horn Syndrome Family. American Journal of Human Genetics, 1997, 61, 233-238.	2.6	33
49	Truncation ofNHEJ1 in a patient with polymicrogyria. Human Mutation, 2007, 28, 356-364.	1.1	33
50	A Kv7.2 mutation associated with early onset epileptic encephalopathy with suppressionâ€burst enhances Kv7/M channel activity. Epilepsia, 2016, 57, e87-93.	2.6	32
51	Early-onset epileptic encephalopathy as the initial clinical presentation of WDR45 deletion in a male patient. European Journal of Human Genetics, 2016, 24, 615-618.	1.4	32
52	Progressive noradrenergic deficits in the locus coeruleus of Mecp2 deficient mice. Journal of Neuroscience Research, 2010, 88, 1500-1509.	1.3	31
53	Alpha-thalassemia/mental retardation syndrome, X-Linked (ATR-X, MIM #301040, ATR-X/XNP/XH2 gene MIM) Tj ETQq1 1 0.784314 rg	1.4	30
54	Severe presentation of WDR62 mutation: Is there a role for modifying genetic factors?. American Journal of Medical Genetics, Part A, 2014, 164, 2161-2171.	0.7	30

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55	Polymorphisms in the C-terminal domain of MECP2 in mentally handicapped boys: implications for genetic counselling. European Journal of Human Genetics, 2002, 10, 86-89.	1.4	29
56	Abnormal function of the UBA5 protein in a case of early developmental and epileptic encephalopathy with suppression-burst. Human Mutation, 2018, 39, 934-938.	1.1	29
57	Clinical study of 19 patients with <scp>SCN</scp>8Aâ€related epilepsy: Two modes of onset regarding <scp>EEG</scp> and seizures. Epilepsia, 2019, 60, 845-856.	2.6	28
58	Severe offtarget effects following intravenous delivery of AAV9-MECP2 in a female mouse model of Rett syndrome. Neurobiology of Disease, 2021, 149, 105235.	2.1	27
59	Intragenic rearrangements in Xâ€linked intellectual deficiency: Results of aâ€CGH in a series of 54 patients and identification of TRPC5 and KLHL15 as potential XLID genes. American Journal of Medical Genetics, Part A, 2014, 164, 1991-1997.	0.7	26
60	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. European Journal of Human Genetics, 2018, 26, 143-148.	1.4	26
61	A knockâ€in mouse model for KCNQ2â€related epileptic encephalopathy displays spontaneous generalized seizures and cognitive impairment. Epilepsia, 2020, 61, 868-878.	2.6	26
62	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. PLoS Biology, 2020, 18, e3000738.	2.6	26
63	Progressive motor and respiratory metabolism deficits in post-weaning Mecp2-null male mice. Behavioural Brain Research, 2011, 216, 313-320.	1.2	24
64	CACNA1A-associated epilepsy: Electroclinical findings and treatment response on seizures in 18 patients. European Journal of Paediatric Neurology, 2021, 33, 75-85.	0.7	24
65	Mapping of X chromosome inversion breakpoints [inv(X)(q11q28)] associated with FG syndrome: A second FG locus [FGS2]?. American Journal of Medical Genetics Part A, 2000, 95, 178-181.	2.4	23
66	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	0.8	23
67	Molecular and clinical descriptions of patients with <scp>GABA_A</scp> receptor gene variants (<scp>GABRA1</scp>, <scp>GABRB2</scp>, <scp>GABRB3</scp>, <scp>GABRG2</scp>): A cohort study, review of literature, and genotypeâ€“phenotype correlation. Epilepsia, 2022, 63, 2519-2533.	2.6	23
68	Map location, genomic organization and expression patterns of the human RED1 RNA editase. Somatic Cell and Molecular Genetics, 1997, 23, 135-145.	0.7	22
69	Rett syndrome from bench to bedside: recent advances. F1000Research, 2018, 7, 398.	0.8	22
70	Huntingtin phosphorylation governs <scp>BDNF</scp> homeostasis and improves the phenotype of Mecp2 knockout mice. EMBO Molecular Medicine, 2020, 12, e10889.	3.3	22
71	Early-onset encephalopathy with paroxysmal movement disorders and epileptic seizures without hemiplegic attacks: About three children with novel ATP1A3 mutations. Brain and Development, 2018, 40, 768-774.	0.6	21
72	Molecular characterization of a new human T-box gene (TBX22) located in Xq21.1 encoding a protein containing a truncated T-domain. Gene, 2000, 255, 289-296.	1.0	19

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73	Rett networked database: An integrated clinical and genetic network of rett syndrome databases. Human Mutation, 2012, 33, 1031-1036.	1.1	19
74	SYNGAP1-DEE: A visual sensitive epilepsy. Clinical Neurophysiology, 2021, 132, 841-850.	0.7	19
75	Ultrasound-Mediated Blood-Brain Barrier Opening Improves Whole Brain Gene Delivery in Mice. Pharmaceutics, 2021, 13, 1245.	2.0	19
76	Construction of a YAC contig spanning the Xq13.3 subband. Genomics, 1995, 26, 115-122.	1.3	18
77	Spectrum of MECP2 Mutations in Rett Syndrome. Genetic Testing and Molecular Biomarkers, 2002, 6, 1-6.	1.7	17
78	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. Gene Expression Patterns, 2009, 9, 423-429.	0.3	17
79	X-linked mental retardation with neonatal hypotonia in a French family (MRX15): Gene assignment to Xp11.22-Xp21.1. , 1996, 64, 97-106.		16
80	Homozygous TBC1D24 mutation in two siblings with familial infantile myoclonic epilepsy (FIME) and moderate intellectual disability. Epilepsy Research, 2015, 111, 72-77.	0.8	16
81	A <scp>N</scp>ovel <scp>H</scp>omozygous <scp>TBC</scp>1<scp>D</scp>24<scp>M</scp>utation <scp>C</scp>ausing <scp>M</scp>ultifocal <scp>M</scp>yoclonus <scp>W</scp>ith <scp>C</scp>erebellar <scp>I</scp>nvolvement. Movement Disorders, 2015, 30, 1431-1432.	2.2	15
82	Effect of desipramine on patients with breathing disorders in <scp>RETT</scp> syndrome. Annals of Clinical and Translational Neurology, 2018, 5, 118-127.	1.7	15
83	Rett syndrome: think outside the (skull) box. Faculty Reviews, 2021, 10, 59.	1.7	15
84	Expression of methyl CpG binding protein 2 (Mecp2) during the postnatal development of the mouse brainstem. Brain Research, 2008, 1236, 176-184.	1.1	12
85	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: Case study and literature review. European Journal of Paediatric Neurology, 2013, 17, 666-670.	0.7	12
86	Heterogeneity of FHF1 related phenotype: Novel case with early onset severe attacks of apnea, partial mitochondrial respiratory chain complex II deficiency, neonatal onset seizures without neurodegeneration. European Journal of Paediatric Neurology, 2017, 21, 783-786.	0.7	12
87	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. Epilepsia, 2020, 61, e71-e78.	2.6	11
88	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	1.1	11
89	Structures, sequence characteristics, and synteny relationships of the transcription factor E4TF1, the splicing factor U2AF35 and the cystathionine beta synthetase genes from Fugu rubripes. Gene, 1999, 226, 211-223.	1.0	10
90	Timeâ€limited alterations in cortical activity of a knockâ€in mouse model of KCNQ2â€related developmental and epileptic encephalopathy. Journal of Physiology, 2022, 600, 2429-2460.	1.3	9

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91	Nhej1 Deficiency Causes Abnormal Development of the Cerebral Cortex. Molecular Neurobiology, 2015, 52, 771-782.	1.9	7
92	Analysis of Astroglial Secretomic Profile in the Mecp2-Deficient Male Mouse Model of Rett Syndrome. International Journal of Molecular Sciences, 2021, 22, 4316.	1.8	7
93	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931.	1.1	7
94	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. , 2011, 155, 880-884.		6
95	A large consanguineous family with a homozygous Metabotropic Glutamate Receptor 7 (mGlu7) variant and developmental epileptic encephalopathy: Effect on protein structure and ligand affinity. Orphanet Journal of Rare Diseases, 2021, 16, 317.	1.2	6
96	Contribution of copy number variants (CNVs) to congenital, unexplained intellectual and developmental disabilities in Lebanese patients. Molecular Cytogenetics, 2015, 8, 26.	0.4	5
97	Patients with KCNH1-related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome. Journal of Medical Genetics, 2022, 59, 505-510.	1.5	5
98	Molecular characterization of a 1p36 chromosomal duplication and in utero interference define ENO1 as a candidate gene for polymicrogyria. European Journal of Human Genetics, 2020, 28, 1703-1713.	1.4	4
99	Exclusion of nine candidate genes for their involvement in X-linked FG syndrome (FGS1) in three families. American Journal of Medical Genetics Part A, 2000, 94, 386-388.	2.4	3
100	Coverage analysis of lists of genes involved in heterogeneous genetic diseases following benchtop exome sequencing using the ion proton. Journal of Genetics, 2016, 95, 203-208.	0.4	3
101	Early-onset epileptic encephalopathy related to germline PIGA mutations: A series of 5 cases. European Journal of Paediatric Neurology, 2020, 28, 214-220.	0.7	3
102	Further delineation of BCAP31-linked intellectual disability: description of 17 new families with LoF and missense variants. European Journal of Human Genetics, 2021, 29, 1405-1417.	1.4	3
103	The EPIGENE network: A French initiative to harmonize and improve the nationwide diagnosis of monogenic epilepsies. European Journal of Medical Genetics, 2022, 65, 104445.	0.7	3
104	Characterization of a de novo balanced translocation in a patient with moderate mental retardation and dysmorphic features. European Journal of Medical Genetics, 2009, 52, 211-217.	0.7	1
105	Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: A distinct MCA/MR syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1147-1151.	0.7	1
106	Overlapping cortical malformations in patients with pathogenic variants inGRIN1andGRIN2B. Journal of Medical Genetics, 2023, 60, 183-192.	1.5	1
107	A polymorphic microsatellite XNP-GT in the XNP/ATRX gene's promotor allows familial indirect diagnosis. Human Mutation, 1999, 14, 448-448.	1.1	0
108	Neurobehavioral Testing of Mouse Models of Rett Syndrome. Neuromethods, 2015, , 399-430.	0.2	0

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109	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0
110	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0
111	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0
112	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0
113	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0
114	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. , 2020, 18, e3000738.		0