

Laurent Villard

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

113
papers

4,224
citations

34
h-index

62
g-index

225
ext. papers

4,902
ext. citations

6.2
avg, IF

4.84
L-index

#	Paper	IF	Citations
113	The EPIGENE network: A French initiative to harmonize and improve the nationwide diagnosis of monogenic epilepsies.. <i>European Journal of Medical Genetics</i> , 2022 , 65, 104445	2.6	0
112	SYNGAP1-DEE: A visual sensitive epilepsy. <i>Clinical Neurophysiology</i> , 2021 , 132, 841-850	4.3	6
111	Analysis of Astroglial Secretomic Profile in the Mecp2-Deficient Male Mouse Model of Rett Syndrome. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	3
110	Patients with -related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome. <i>Journal of Medical Genetics</i> , 2021 ,	5.8	1
109	Rett syndrome: think outside the (skull) box. <i>Faculty Reviews</i> , 2021 , 10, 59	1.2	1
108	A large consanguineous family with a homozygous Metabotropic Glutamate Receptor 7 (mGlu7) variant and developmental epileptic encephalopathy: Effect on protein structure and ligand affinity. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 317	4.2	0
107	CACNA1A-associated epilepsy: Electroclinical findings and treatment response on seizures in 18 patients. <i>European Journal of Paediatric Neurology</i> , 2021 , 33, 75-85	3.8	5
106	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. <i>Genetics in Medicine</i> , 2021 , 23, 2138-2149	8.1	1
105	Severe offtarget effects following intravenous delivery of AAV9-MECP2 in a female mouse model of Rett syndrome. <i>Neurobiology of Disease</i> , 2021 , 149, 105235	7.5	9
104	Further delineation of BCAP31-linked intellectual disability: description of 17 new families with LoF and missense variants. <i>European Journal of Human Genetics</i> , 2021 , 29, 1405-1417	5.3	0
103	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. <i>Brain Sciences</i> , 2021 , 11,	3.4	2
102	Ultrasound-Mediated Blood-Brain Barrier Opening Improves Whole Brain Gene Delivery in Mice. <i>Pharmaceutics</i> , 2021 , 13,	6.4	3
101	Molecular characterization of a 1p36 chromosomal duplication and in utero interference define ENO1 as a candidate gene for polymicrogyria. <i>European Journal of Human Genetics</i> , 2020 , 28, 1703-1713	5.3	1
100	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2020 , 61, e71-e78	6.8	2
99	A knock-in mouse model for KCNQ2-related epileptic encephalopathy displays spontaneous generalized seizures and cognitive impairment. <i>Epilepsia</i> , 2020 , 61, 868-878	6.4	13
98	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. <i>PLoS Biology</i> , 2020 , 18, e3000738	9.7	7
97	Huntingtin phosphorylation governs BDNF homeostasis and improves the phenotype of Mecp2 knockout mice. <i>EMBO Molecular Medicine</i> , 2020 , 12, e10889	12	14

96	Early-onset epileptic encephalopathy related to germline PIGA mutations: A series of 5 cases. <i>European Journal of Paediatric Neurology</i> , 2020 , 28, 214-220	3.8	1
95	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
94	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
93	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
92	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
91	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
90	The M-current works in tandem with the persistent sodium current to set the speed of locomotion 2020 , 18, e3000738		
89	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019 , 104, 1210-1222	11	31
88	Clinical study of 19 patients with SCN8A-related epilepsy: Two modes of onset regarding EEG and seizures. <i>Epilepsia</i> , 2019 , 60, 845-856	6.4	20
87	Analysis of the Phenotypes in the Rett Networked Database. <i>International Journal of Genomics</i> , 2019 , 2019, 6956934	2.5	14
86	Exome sequencing in congenital ataxia identifies two new candidate genes and highlights a pathophysiological link between some congenital ataxias and early infantile epileptic encephalopathies. <i>Genetics in Medicine</i> , 2019 , 21, 553-563	8.1	25
85	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , 2019 , 21, 837-849	8.1	32
84	The phenotypic spectrum of WWOX-related disorders: 20 additional cases of WOREE syndrome and review of the literature. <i>Genetics in Medicine</i> , 2019 , 21, 1308-1318	8.1	22
83	Effect of desipramine on patients with breathing disorders in RETT syndrome. <i>Annals of Clinical and Translational Neurology</i> , 2018 , 5, 118-127	5.3	14
82	Abnormal function of the UBA5 protein in a case of early developmental and epileptic encephalopathy with suppression-burst. <i>Human Mutation</i> , 2018 , 39, 934-938	4.7	18
81	Early-onset encephalopathy with paroxysmal movement disorders and epileptic seizures without hemiplegic attacks: About three children with novel ATP1A3 mutations. <i>Brain and Development</i> , 2018 , 40, 768-774	2.2	14
80	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. <i>European Journal of Human Genetics</i> , 2018 , 26, 143-148	5.3	15
79	Rett syndrome from bench to bedside: recent advances. <i>F1000Research</i> , 2018 , 7, 398	3.6	13

78	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). <i>Human Genetics</i> , 2018 , 137, 753-768	6.3	19
77	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. <i>European Journal of Paediatric Neurology</i> , 2017 , 21, 783-786	3.8	9
76	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. <i>Brain</i> , 2017 , 140, 1316-1336	11.2	285
75	A codon-optimized Mecp2 transgene corrects breathing deficits and improves survival in a mouse model of Rett syndrome. <i>Neurobiology of Disease</i> , 2017 , 99, 1-11	7.5	22
74	Early-onset epileptic encephalopathy as the initial clinical presentation of WDR45 deletion in a male patient. <i>European Journal of Human Genetics</i> , 2016 , 24, 615-8	5.3	27
73	A Kv7.2 mutation associated with early onset epileptic encephalopathy with suppression-burst enhances Kv7/M channel activity. <i>Epilepsia</i> , 2016 , 57, e87-93	6.4	19
72	Coverage Analysis of Lists of Genes Involved in Heterogeneous Genetic Diseases following Benchtop Exome Sequencing using the Ion Proton. <i>Journal of Genetics</i> , 2016 , 95, 203-8	1.2	3
71	Clinical and molecular findings in 39 patients with KBG syndrome caused by deletion or mutation of ANKRD11. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2847-2859	2.5	39
70	Nhej1 Deficiency Causes Abnormal Development of the Cerebral Cortex. <i>Molecular Neurobiology</i> , 2015 , 52, 771-82	6.2	5
69	Epilepsy in Rett syndrome--lessons from the Rett networked database. <i>Epilepsia</i> , 2015 , 56, 569-76	6.4	30
68	Evidence that homozygous PTPRD gene microdeletion causes trigonocephaly, hearing loss, and intellectual disability. <i>Molecular Cytogenetics</i> , 2015 , 8, 39	2	27
67	Contribution of copy number variants (CNVs) to congenital, unexplained intellectual and developmental disabilities in Lebanese patients. <i>Molecular Cytogenetics</i> , 2015 , 8, 26	2	3
66	Variable clinical expression in patients with mosaicism for KCNQ2 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2314-8	2.5	27
65	Epileptic patients with de novo STXBP1 mutations: Key clinical features based on 24 cases. <i>Epilepsia</i> , 2015 , 56, 1931-40	6.4	31
64	A Novel Homozygous TBC1D24 Mutation Causing Multifocal Myoclonus With Cerebellar Involvement. <i>Movement Disorders</i> , 2015 , 30, 1431-2	7	13
63	A recurrent KCNQ2 pore mutation causing early onset epileptic encephalopathy has a moderate effect on M current but alters subcellular localization of Kv7 channels. <i>Neurobiology of Disease</i> , 2015 , 80, 80-92	7.5	45
62	Homozygous TBC1D24 mutation in two siblings with familial infantile myoclonic epilepsy (FIME) and moderate intellectual disability. <i>Epilepsy Research</i> , 2015 , 111, 72-7	3	14
61	Neurobehavioral Testing of Mouse Models of Rett Syndrome. <i>Neuromethods</i> , 2015 , 399-430	0.4	

60	Intragenic rearrangements in X-linked intellectual deficiency: results of a-CGH in a series of 54 patients and identification of TRPC5 and KLHL15 as potential XLID genes. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1991-7	2.5	16
59	Mutations in SLC13A5 cause autosomal-recessive epileptic encephalopathy with seizure onset in the first days of life. <i>American Journal of Human Genetics</i> , 2014 , 95, 113-20	11	80
58	GABA and glutamate pathways are spatially and developmentally affected in the brain of Mecp2-deficient mice. <i>PLoS ONE</i> , 2014 , 9, e92169	3.7	41
57	AP1S2 is mutated in X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). <i>European Journal of Human Genetics</i> , 2014 , 22, 363-8	5.3	36
56	Severe presentation of WDR62 mutation: is there a role for modifying genetic factors?. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2161-71	2.5	26
55	Similar early characteristics but variable neurological outcome of patients with a de novo mutation of KCNQ2. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 80	4.2	64
54	Mutations in BCAP31 cause a severe X-linked phenotype with deafness, dystonia, and central hypomyelination and disorganize the Golgi apparatus. <i>American Journal of Human Genetics</i> , 2013 , 93, 579-86	11	41
53	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: case study and literature review. <i>European Journal of Paediatric Neurology</i> , 2013 , 17, 666-70	3.8	11
52	Novel compound heterozygous mutations in TBC1D24 cause familial malignant migrating partial seizures of infancy. <i>Human Mutation</i> , 2013 , 34, 869-72	4.7	99
51	Loss of function of KIAA2022 causes mild to severe intellectual disability with an autism spectrum disorder and impairs neurite outgrowth. <i>Human Molecular Genetics</i> , 2013 , 22, 3306-14	5.6	45
50	Modification of Mecp2 dosage alters axonal transport through the Huntingtin/Hap1 pathway. <i>Neurobiology of Disease</i> , 2012 , 45, 786-95	7.5	54
49	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. <i>Human Mutation</i> , 2012 , 33, 1031-6	4.7	14
48	Progressive motor and respiratory metabolism deficits in post-weaning Mecp2-null male mice. <i>Behavioural Brain Research</i> , 2011 , 216, 313-20	3.4	20
47	Epileptic and nonepileptic features in patients with early onset epileptic encephalopathy and STXBP1 mutations. <i>Epilepsia</i> , 2011 , 52, 1828-34	6.4	90
46	Morphological and functional alterations in the substantia nigra pars compacta of the Mecp2-null mouse. <i>Neurobiology of Disease</i> , 2011 , 41, 385-97	7.5	39
45	Biogenic amines and their metabolites are differentially affected in the Mecp2-deficient mouse brain. <i>BMC Neuroscience</i> , 2011 , 12, 47	3.2	28
44	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 880-4	2.5	6
43	Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: a distinct MCA/MR syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1147-51	2.5	1

42	Disruption of the ATP8A2 gene in a patient with a t(10;13) de novo balanced translocation and a severe neurological phenotype. <i>European Journal of Human Genetics</i> , 2010 , 18, 1360-3	5.3	48
41	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. <i>Nature Genetics</i> , 2010 , 42, 1021-6	36.3	347
40	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. <i>Journal of Medical Genetics</i> , 2010 , 47, 155-61	5.8	45
39	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. <i>Brain</i> , 2010 , 133, 3194-209	11.2	108
38	Deletion of YWHAE in a patient with periventricular heterotopias and pronounced corpus callosum hypoplasia. <i>Journal of Medical Genetics</i> , 2010 , 47, 132-6	5.8	32
37	Progressive noradrenergic deficits in the locus coeruleus of Mecp2 deficient mice. <i>Journal of Neuroscience Research</i> , 2010 , 88, 1500-9	4.4	30
36	Biogenic amines in Rett syndrome: the usual suspects. <i>Behavior Genetics</i> , 2010 , 40, 59-75	3.2	27
35	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. <i>Gene Expression Patterns</i> , 2009 , 9, 423-9	1.5	15
34	Characterization of a de novo balanced translocation in a patient with moderate mental retardation and dysmorphic features. <i>European Journal of Medical Genetics</i> , 2009 , 52, 211-7	2.6	1
33	Expression of methyl CpG binding protein 2 (Mecp2) during the postnatal development of the mouse brainstem. <i>Brain Research</i> , 2008 , 1236, 176-84	3.7	11
32	Tyrosine hydroxylase deficit in the chemoafferent and the sympathoadrenergic pathways of the Mecp2 deficient mouse. <i>Neuroscience Letters</i> , 2008 , 447, 82-6	3.3	42
31	TCF4 deletions in Pitt-Hopkins Syndrome. <i>Human Mutation</i> , 2008 , 29, E242-51	4.7	47
30	Metabolic fingerprints of altered brain growth, osmoregulation and neurotransmission in a Rett syndrome model. <i>PLoS ONE</i> , 2007 , 2, e157	3.7	38
29	Truncation of NHEJ1 in a patient with polymicrogyria. <i>Human Mutation</i> , 2007 , 28, 356-64	4.7	32
28	A cluster of translocation breakpoints in 2q37 is associated with overexpression of NPPC in patients with a similar overgrowth phenotype. <i>Human Mutation</i> , 2007 , 28, 1183-8	4.7	72
27	Treatment with desipramine improves breathing and survival in a mouse model for Rett syndrome. <i>European Journal of Neuroscience</i> , 2007 , 25, 1915-22	3.5	110
26	MECP2 mutations in males. <i>Journal of Medical Genetics</i> , 2007 , 44, 417-23	5.8	120
25	Brain magnetic resonance study of Mecp2 deletion effects on anatomy and metabolism. <i>Biochemical and Biophysical Research Communications</i> , 2006 , 340, 776-83	3.4	62

24	The incidence of Rett syndrome in France. <i>Pediatric Neurology</i> , 2006 , 34, 372-5	2.9	35
23	Mecp2 deficiency disrupts norepinephrine and respiratory systems in mice. <i>Journal of Neuroscience</i> , 2005 , 25, 11521-30	6.6	222
22	Spectrum of MECP2 mutations in Rett syndrome. <i>Genetic Testing and Molecular Biomarkers</i> , 2002 , 6, 1-6		16
21	Craniofacial expression of human and murine TBX22 correlates with the cleft palate and ankyloglossia phenotype observed in CPX patients. <i>Human Molecular Genetics</i> , 2002 , 11, 2793-804	5.6	76
20	Alpha-thalassemia/mental retardation syndrome, X-Linked (ATR-X, MIM #301040, ATR-X/XNP/XH2 gene MIM #300032). <i>European Journal of Human Genetics</i> , 2002 , 10, 223-5	5.3	25
19	Polymorphisms in the C-terminal domain of MECP2 in mentally handicapped boys: implications for genetic counselling. <i>European Journal of Human Genetics</i> , 2002 , 10, 86-9	5.3	27
18	A locus for bilateral perisylvian polymicrogyria maps to Xq28. <i>American Journal of Human Genetics</i> , 2002 , 70, 1003-8	11	97
17	Identification of a mutation in the XNP/ATR-X gene in a family reported as Smith-Fineman-Myers syndrome 2000 , 91, 83-85		34
16	Exclusion of nine candidate genes for their involvement in X-linked FG syndrome (FGS1) in three families. <i>American Journal of Medical Genetics Part A</i> , 2000 , 94, 386-8		3
15	Mapping of X chromosome inversion breakpoints [inv(X)(q11q28)] associated with FG syndrome: a second FG locus [FGS2]?. <i>American Journal of Medical Genetics Part A</i> , 2000 , 95, 178-81		22
14	Linkage of X-linked myopathy with excessive autophagy (XMEA) to Xq28. <i>European Journal of Human Genetics</i> , 2000 , 8, 125-9	5.3	38
13	Molecular characterization of a new human T-box gene (TBX22) located in xq21.1 encoding a protein containing a truncated T-domain. <i>Gene</i> , 2000 , 255, 289-96	3.8	18
12	A polymorphic microsatellite XNP-GT in the XNP/ATRX gene promoter allows familial indirect diagnosis. <i>Human Mutation</i> , 1999 , 14, 448	4.7	
11	Carpenter-Waziri syndrome results from a mutation in XNP. <i>American Journal of Medical Genetics Part A</i> , 1999 , 85, 249-51		40
10	Structures, sequence characteristics, and synteny relationships of the transcription factor E4TF1, the splicing factor U2AF35 and the cystathionine beta synthetase genes from <i>Fugu rubripes</i> . <i>Gene</i> , 1999 , 226, 211-23	3.8	10
9	Determination of the genomic structure of the XNP/ATRX gene encoding a potential zinc finger helicase. <i>Genomics</i> , 1997 , 43, 149-55	4.3	57
8	A C2055T transition in exon 8 of the ATP7A gene is associated with exon skipping in an occipital horn syndrome family. <i>American Journal of Human Genetics</i> , 1997 , 61, 233-8	11	30
7	Map location, genomic organization and expression patterns of the human RED1 RNA editase. <i>Somatic Cell and Molecular Genetics</i> , 1997 , 23, 135-45		19

6	X-linked mental retardation with neonatal hypotonia in a French family (MRX15): gene assignment to Xp11.22-Xp21.1. <i>American Journal of Medical Genetics Part A</i> , 1996 , 64, 97-106		12
5	XNP mutation in a large family with Juberg-Marsidi syndrome. <i>Nature Genetics</i> , 1996 , 12, 359-60	36.3	90
4	Construction of a YAC contig spanning the Xq13.3 subband. <i>Genomics</i> , 1995 , 26, 115-22	4.3	15
3	Mutations in a putative global transcriptional regulator cause X-linked mental retardation with alpha-thalassemia (ATR-X syndrome). <i>Cell</i> , 1995 , 80, 837-45	56.2	511
2	It takes two to tango: M-current swings with the persistent sodium current to set the speed of locomotion		1
1	Time-limited alterations in cortical activity of a Knock-in mice model of KCNQ2-related Developmental and Epileptic Encephalopathy		1