Laurent Villard

List of Publications by Citations

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	Mutations in a putative global transcriptional regulator cause X-linked mental retardation with alpha-thalassemia (ATR-X syndrome). *Cell*, **1995**, 80, 837-45	56.2	511
112	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. *Nature Genetics*, **2010**, 42, 1021-6	36.3	347
111	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. *Brain*, **2017**, 140, 1316-1336	11.2	285
110	Mecp2 deficiency disrupts norepinephrine and respiratory systems in mice. *Journal of Neuroscience*, **2005**, 25, 11521-30	6.6	222
109	MECP2 mutations in males. *Journal of Medical Genetics*, **2007**, 44, 417-23	5.8	120
108	Treatment with desipramine improves breathing and survival in a mouse model for Rett syndrome. *European Journal of Neuroscience*, **2007**, 25, 1915-22	3.5	110
107	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. *Brain*, **2010**, 133, 3194-209	11.2	108
106	Novel compound heterozygous mutations in TBC1D24 cause familial malignant migrating partial seizures of infancy. *Human Mutation*, **2013**, 34, 869-72	4.7	99
105	A locus for bilateral perisylvian polymicrogyria maps to Xq28. *American Journal of Human Genetics*, **2002**, 70, 1003-8	11	97
104	Epileptic and nonepileptic features in patients with early onset epileptic encephalopathy and STXBP1 mutations. *Epilepsia*, **2011**, 52, 1828-34	6.4	90
103	XNP mutation in a large family with Juberg-Marsidi syndrome. *Nature Genetics*, **1996**, 12, 359-60	36.3	90
102	Mutations in SLC13A5 cause autosomal-recessive epileptic encephalopathy with seizure onset in the first days of life. *American Journal of Human Genetics*, **2014**, 95, 113-20	11	80
101	Craniofacial expression of human and murine TBX22 correlates with the cleft palate and ankyloglossia phenotype observed in CPX patients. *Human Molecular Genetics*, **2002**, 11, 2793-804	5.6	76
100	A cluster of translocation breakpoints in 2q37 is associated with overexpression of NPPC in patients with a similar overgrowth phenotype. *Human Mutation*, **2007**, 28, 1183-8	4.7	72
99	Similar early characteristics but variable neurological outcome of patients with a de novo mutation of KCNQ2. *Orphanet Journal of Rare Diseases*, **2013**, 8, 80	4.2	64
98	Brain magnetic resonance study of Mecp2 deletion effects on anatomy and metabolism. *Biochemical and Biophysical Research Communications*, **2006**, 340, 776-83	3.4	62
97	Determination of the genomic structure of the XNP/ATRX gene encoding a potential zinc finger helicase. *Genomics*, **1997**, 43, 149-55	4.3	57

96	Modification of Mecp2 dosage alters axonal transport through the Huntingtin/Hap1 pathway. *Neurobiology of Disease*, **2012**, 45, 786-95	7.5	54
95	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, 1360-3	5.3	48
94	TCF4 deletions in Pitt-Hopkins Syndrome. *Human Mutation*, **2008**, 29, E242-51	4.7	47
93	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	7.5	45
92	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, 3306-14	5.6	45
91	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. *Journal of Medical Genetics*, **2010**, 47, 155-61	5.8	45
90	Tyrosine hydroxylase deficit in the chemoafferent and the sympathoadrenergic pathways of the Mecp2 deficient mouse. *Neuroscience Letters*, **2008**, 447, 82-6	3.3	42
89	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, 579-86	11	41
88	GABA and glutamate pathways are spatially and developmentally affected in the brain of Mecp2-deficient mice. *PLoS ONE*, **2014**, 9, e92169	3.7	41
87	Carpenter-Waziri syndrome results from a mutation in XNP. *American Journal of Medical Genetics Part A*, **1999**, 85, 249-51		40
86	Morphological and functional alterations in the substantia nigra pars compacta of the Mecp2-null mouse. *Neurobiology of Disease*, **2011**, 41, 385-97	7.5	39
85	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	2.5	39
84	Metabolic fingerprints of altered brain growth, osmoregulation and neurotransmission in a Rett syndrome model. *PLoS ONE*, **2007**, 2, e157	3.7	38
83	Linkage of X-linked myopathy with excessive autophagy (XMEA) to Xq28. *European Journal of Human Genetics*, **2000**, 8, 125-9	5.3	38
82	AP1S2 is mutated in X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). *European Journal of Human Genetics*, **2014**, 22, 363-8	5.3	36
81	The incidence of Rett syndrome in France. *Pediatric Neurology*, **2006**, 34, 372-5	2.9	35
80	Identification of a mutation in the XNP/ATR-X gene in a family reported as Smith-Fineman-Myers syndrome **2000**, 91, 83-85		34
79	Deletion of YWHAE in a patient with periventricular heterotopias and pronounced corpus callosum hypoplasia. *Journal of Medical Genetics*, **2010**, 47, 132-6	5.8	32

78	Truncation of NHEJ1 in a patient with polymicrogyria. *Human Mutation*, **2007**, 28, 356-64	4.7	32
77	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
76	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
75	Epileptic patients with de novo STXBP1 mutations: Key clinical features based on 24 cases. *Epilepsia* , **2015**, 56, 1931-40	6.4	31
74	Epilepsy in Rett syndrome--lessons from the Rett networked database. *Epilepsia*, **2015**, 56, 569-76	6.4	30
73	Progressive noradrenergic deficits in the locus coeruleus of Mecp2 deficient mice. *Journal of Neuroscience Research*, **2010**, 88, 1500-9	4.4	30
72	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, 233-8	11	30
71	Biogenic amines and their metabolites are differentially affected in the Mecp2-deficient mouse brain. *BMC Neuroscience*, **2011**, 12, 47	3.2	28
70	Early-onset epileptic encephalopathy as the initial clinical presentation of WDR45 deletion in a male patient. *European Journal of Human Genetics*, **2016**, 24, 615-8	5.3	27
69	Evidence that homozygous PTPRD gene microdeletion causes trigonocephaly, hearing loss, and intellectual disability. *Molecular Cytogenetics*, **2015**, 8, 39	2	27
68	Variable clinical expression in patients with mosaicism for KCNQ2 mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2314-8	2.5	27
67	Biogenic amines in Rett syndrome: the usual suspects. *Behavior Genetics*, **2010**, 40, 59-75	3.2	27
66	Polymorphisms in the C-terminal domain of MECP2 in mentally handicapped boys: implications for genetic counselling. *European Journal of Human Genetics*, **2002**, 10, 86-9	5.3	27
65	Severe presentation of WDR62 mutation: is there a role for modifying genetic factors?. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2161-71	2.5	26
64	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	8.1	25
63	Alpha-thalassemia/mental retardation syndrome, X-Linked (ATR-X, MIM #301040, ATR-X/XNP/XH2 gene MIM #300032). *European Journal of Human Genetics*, **2002**, 10, 223-5	5.3	25
62	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	7.5	22
61	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, 178-81		22

60	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	8.1	22
59	Clinical study of 19 patients with SCN8A-related epilepsy: Two modes of onset regarding EEG and seizures. *Epilepsia*, **2019**, 60, 845-856	6.4	20
58	Progressive motor and respiratory metabolism deficits in post-weaning Mecp2-null male mice. *Behavioural Brain Research*, **2011**, 216, 313-20	3.4	20
57	Map location, genomic organization and expression patterns of the human RED1 RNA editase. *Somatic Cell and Molecular Genetics*, **1997**, 23, 135-45		19
56	A Kv7.2 mutation associated with early onset epileptic encephalopathy with suppression-burst enhances Kv7/M channel activity. *Epilepsia*, **2016**, 57, e87-93	6.4	19
55	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). *Human Genetics*, **2018**, 137, 753-768	6.3	19
54	Abnormal function of the UBA5 protein in a case of early developmental and epileptic encephalopathy with suppression-burst. *Human Mutation*, **2018**, 39, 934-938	4.7	18
53	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, 289-96	3.8	18
52	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. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1991-7	2.5	16
51	Spectrum of MECP2 mutations in Rett syndrome. *Genetic Testing and Molecular Biomarkers*, **2002**, 6, 1-6		16
50	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. *Gene Expression Patterns*, **2009**, 9, 423-9	1.5	15
49	Construction of a YAC contig spanning the Xq13.3 subband. *Genomics*, **1995**, 26, 115-22	4.3	15
48	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	5.3	15
47	Analysis of the Phenotypes in the Rett Networked Database. *International Journal of Genomics*, **2019**, 2019, 6956934	2.5	14
46	Effect of desipramine on patients with breathing disorders in RETT syndrome. *Annals of Clinical and Translational Neurology*, **2018**, 5, 118-127	5.3	14
45	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	2.2	14
44	Homozygous TBC1D24 mutation in two siblings with familial infantile myoclonic epilepsy (FIME) and moderate intellectual disability. *Epilepsy Research*, **2015**, 111, 72-7	3	14
43	Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. *Human Mutation*, **2012**, 33, 1031-6	4.7	14

42	Huntingtin phosphorylation governs BDNF homeostasis and improves the phenotype of Mecp2 knockout mice. *EMBO Molecular Medicine*, **2020**, 12, e10889	12	14
41	A knock-in mouse model for KCNQ2-related epileptic encephalopathy displays spontaneous generalized seizures and cognitive impairment. *Epilepsia*, **2020**, 61, 868-878	6.4	13
40	A Novel Homozygous TBC1D24 Mutation Causing Multifocal Myoclonus With Cerebellar Involvement. *Movement Disorders*, **2015**, 30, 1431-2	7	13
39	Rett syndrome from bench to bedside: recent advances. *F1000Research*, **2018**, 7, 398	3.6	13
38	X-linked mental retardation with neonatal hypotonia in a French family (MRX15): gene assignment to Xp11.22-Xp21.1. *American Journal of Medical Genetics Part A*, **1996**, 64, 97-106		12
37	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: case study and literature review. *European Journal of Paediatric Neurology*, **2013**, 17, 666-70	3.8	11
36	Expression of methyl CpG binding protein 2 (Mecp2) during the postnatal development of the mouse brainstem. *Brain Research*, **2008**, 1236, 176-84	3.7	11
35	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, 211-23	3.8	10
34	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	3.8	9
33	Severe offtarget effects following intravenous delivery of AAV9-MECP2 in a female mouse model of Rett syndrome. *Neurobiology of Disease*, **2021**, 149, 105235	7.5	9
32	The M-current works in tandem with the persistent sodium current to set the speed of locomotion. *PLoS Biology*, **2020**, 18, e3000738	9.7	7
31	How many entities exist for the spectrum of disorders associated with brachydactyly, syndactyly, short stature, microcephaly, and intellectual disability?. *American Journal of Medical Genetics, Part A* , **2011**, 155A, 880-4	2.5	6
30	SYNGAP1-DEE: A visual sensitive epilepsy. *Clinical Neurophysiology*, **2021**, 132, 841-850	4.3	6
29	Nhej1 Deficiency Causes Abnormal Development of the Cerebral Cortex. *Molecular Neurobiology*, **2015**, 52, 771-82	6.2	5
28	CACNA1A-associated epilepsy: Electroclinical findings and treatment response on seizures in 18 patients. *European Journal of Paediatric Neurology*, **2021**, 33, 75-85	3.8	5
27	Contribution of copy number variants (CNVs) to congenital, unexplained intellectual and developmental disabilities in Lebanese patients. *Molecular Cytogenetics*, **2015**, 8, 26	2	3
26	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, 386-8		3
25	Analysis of Astroglial Secretomic Profile in the Mecp2-Deficient Male Mouse Model of Rett Syndrome. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3

24	Coverage Analysis of Lists of Genes involved in Heterogeneous Genetic Diseases following Benchtop Exome Sequencing using the Ion Proton. *Journal of Genetics*, **2016**, 95, 203-8	1.2	3
23	Ultrasound-Mediated Blood-Brain Barrier Opening Improves Whole Brain Gene Delivery in Mice. *Pharmaceutics*, **2021**, 13,	6.4	3
22	Defining the phenotype of FHF1 developmental and epileptic encephalopathy. *Epilepsia*, **2020**, 61, e71-e78	6.4	2
21	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. *Brain Sciences*, **2021**, 11,	3.4	2
20	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	5.3	1
19	Ambiguous genitalia, microcephaly, seizures, bone malformations, and early death: a distinct MCA/MR syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1147-51	2.5	1
18	Characterization of a de novo balanced translocation in a patient with moderate mental retardation and dysmorphic features. *European Journal of Medical Genetics*, **2009**, 52, 211-7	2.6	1
17	It takes two to tango: M-current swings with the persistent sodium current to set the speed of locomotion		1
16	Time-limited alterations in cortical activity of a Knock-in mice model of KCNQ2-related Developmental and Epileptic Encephalopathy		1
15	Early-onset epileptic encephalopathy related to germline PIGA mutations: A series of 5 cases. *European Journal of Paediatric Neurology*, **2020**, 28, 214-220	3.8	1
14	Patients with -related intellectual disability without distinctive features of Zimmermann-Laband/Temple-Baraitser syndrome. *Journal of Medical Genetics*, **2021**,	5.8	1
13	Rett syndrome: think outside the (skull) box. *Faculty Reviews*, **2021**, 10, 59	1.2	1
12	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. *Genetics in Medicine*, **2021**, 23, 2138-2149	8.1	1
11	The EPIGENE network: A French initiative to harmonize and improve the nationwide diagnosis of monogenic epilepsies.. *European Journal of Medical Genetics*, **2022**, 65, 104445	2.6	0
10	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	4.2	0
9	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	5.3	0
8	A polymorphic microsatellite XNP-GT in the XNP/ATRX gene promotor allows familial indirect diagnosis. *Human Mutation*, **1999**, 14, 448	4.7	
7	Neurobehavioral Testing of Mouse Models of Rett Syndrome. *Neuromethods*, **2015**, 399-430	0.4	

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