Laurence Colleaux

List of Publications by Year
in descending order

85	6,069	66336	74160
papers	citations	42 h-index	75 g-index

90	90	90	11044
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
2	MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087.	12.8	28
3	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30
4	Loss of the neurodevelopmental disease-associated gene miR-146a impairs neural progenitor differentiation and causes learning and memory deficits. Molecular Autism, 2020, 11, 22.	4.9	24
5	Exome sequencing findings in 27 patients with myoclonicâ€atonic epilepsy: Is there a major genetic factor?. Clinical Genetics, 2019, 96, 254-260.	2.0	25
6	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
7	Whole-exome sequence analysis highlights the role of unmasked recessive mutations in copy number variants with incomplete penetrance. European Journal of Human Genetics, 2018, 26, 912-918.	2.8	8
8	Novel de novo ZBTB20 mutations in three cases with Primrose syndrome and constant corpus callosum anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 1091-1098.	1.2	16
9	NONO Detects the Nuclear HIV Capsid to Promote cGAS-Mediated Innate Immune Activation. Cell, 2018, 175, 488-501.e22.	28.9	154
10	Distal duplication of chromosome 16q22.1q23.1 in a Vietnamese patient with midface hypoplasia and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 1981-1984.	1.2	2
11	Role of miR-146a in neural stem cell differentiation and neural lineage determination: relevance for neurodevelopmental disorders. Molecular Autism, 2018, 9, 38.	4.9	70
12	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	7.6	67
13	High N-glycan multiplicity is critical for neuronal adhesion and sensitizes the developing cerebellum to N-glycosylation defect. ELife, 2018, 7, .	6.0	43
14	AMPA-receptor specific biogenesis complexes control synaptic transmission and intellectual ability. Nature Communications, 2017, 8, 15910.	12.8	77
15	Refining the phenotype associated with CASC5 mutation. Neurogenetics, 2016, 17, 71-78.	1.4	21
16	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	6.2	50
17	The emerging roles of MicroRNAs in autism spectrum disorders. Neuroscience and Biobehavioral Reviews, 2016, 71, 729-738.	6.1	51
18	Utility of whole exome sequencing for the early diagnosis of pediatric-onset cerebellar atrophy associated with developmental delay in an inbred population. Orphanet Journal of Rare Diseases, 2016, 11, 57.	2.7	31

#	Article	IF	Citations
19	Profiling olfactory stem cells from living patients identifies miRNAs relevant for autism pathophysiology. Molecular Autism, 2016, 7, 1.	4.9	114
20	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
21	A nonsense variant in HERC1 is associated with intellectual disability, megalencephaly, thick corpus callosum and cerebellar atrophy. European Journal of Human Genetics, 2016, 24, 455-458.	2.8	53
22	Contiguous mutation syndrome in the era of highâ€throughput sequencing. Molecular Genetics & Genomic Medicine, 2015, 3, 215-220.	1.2	9
23	Mutations in NONO lead to syndromic intellectual disability and inhibitory synaptic defects. Nature Neuroscience, 2015, 18, 1731-1736.	14.8	65
24	Phenotypeâ€“genotype correlations in 17 new patients with an Xp11.23p11.22 microduplication and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 111-122.	1.2	23
25	Human Slack Potassium Channel Mutations Increase Positive Cooperativity between Individual Channels. Cell Reports, 2014, 9, 1661-1672.	6.4	97
26	Multiple congenital anomaliesâ€intellectual disability (MCAâ€ID) and neuroblastoma in a patient harboring a de novo 14q23.1q23.3 deletion. American Journal of Medical Genetics, Part A, 2014, 164, 1310-1317.	1.2	9
27	Mutations in QARS, Encoding Glutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. American Journal of Human Genetics, 2014, 94, 547-558.	6.2	106
28	Deficiency of Asparagine Synthetase Causes Congenital Microcephaly and a Progressive Form of Encephalopathy. Neuron, 2013, 80, 429-441.	8.1	137
29	Use of Resonance-Wavelength Grating Optical Biosensors to Detect Channel-Protein Interaction in Slack KNa Channels. Biophysical Journal, 2013, 104, 131a.	0.5	0
30	NF-ÎºB signalling requirement for brain myelin formation is shown by genotype/MRI phenotype correlations in patients with Xq28 duplications. European Journal of Human Genetics, 2013, 21, 195-199.	2.8	21
31	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. Journal of Experimental Medicine, 2013, 210, 2503-2513.	8.5	33
32	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.	2.9	62
33	Xq25 duplications encompassing <scp>GRIA</scp>3 and <scp>STAG</scp>2 genes in two families convey recognizable Xâ€linked intellectual disability with distinctive facial appearance. American Journal of Medical Genetics, Part A, 2013, 161, 1370-1375.	1.2	25
34	Mutation in TTI2 Reveals a Role for Triple T Complex in Human Brain Development. Human Mutation, 2013, 34, 1472-1476.	2.5	25
35	De novo gain-of-function KCNT1 channel mutations cause malignant migrating partial seizures of infancy. Nature Genetics, 2012, 44, 1255-1259.	21.4	436
36	eIF2Î³ Mutation that Disrupts eIF2 Complex Integrity Links Intellectual Disability to Impaired Translation Initiation. Molecular Cell, 2012, 48, 641-646.	9.7	63

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37	MED23 Mutation Links Intellectual Disability to Dysregulation of Immediate Early Gene Expression. Science, 2011, 333, 1161-1163.	12.6	106
38	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. American Journal of Human Genetics, 2011, 88, 788-795.	6.2	206
39	Distinct Effects of Allelic NFIX Mutations on Nonsense-Mediated mRNA Decay Engender Either a Sotos-like or a Marshall-Smith Syndrome. American Journal of Human Genetics, 2010, 87, 189-198.	6.2	131
40	SOBP Is Mutated in Syndromic and Nonsyndromic Intellectual Disability and Is Highly Expressed in the Brain Limbic System. American Journal of Human Genetics, 2010, 87, 694-700.	6.2	20
41	Familial interstitial Xq27.3q28 duplication encompassing the FMR1 gene but not the MECP2 gene causes a new syndromic mental retardation condition. European Journal of Human Genetics, 2010, 18, 285-290.	2.8	31
42	Array-based comparative genomic hybridization identifies a high frequency of copy number variations in patients with syndromic overgrowth. European Journal of Human Genetics, 2010, 18, 227-232.	2.8	20
43	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
44	Genotypeâ€“phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21. European Journal of Human Genetics, 2009, 17, 454-466.	2.8	240
45	Loss-of-Function Mutation in the Dioxygenase-Encoding FTO Gene Causes Severe Growth Retardation and Multiple Malformations. American Journal of Human Genetics, 2009, 85, 106-111.	6.2	340
46	Combination of Linkage Mapping and Microarray-Expression Analysis Identifies NF-ÎºB Signaling Defect as a Cause of Autosomal-Recessive Mental Retardation. American Journal of Human Genetics, 2009, 85, 903-908.	6.2	96
47	Compound heterozygous ASPM mutations associated with microcephaly and simplified cortical gyration in a consanguineous Algerian family. European Journal of Medical Genetics, 2009, 52, 180-184.	1.3	25
48	Clinical, cellular, and neuropathological consequences ofAP1S2mutations: further delineation of a recognizable X-linked mental retardation syndrome. Human Mutation, 2008, 29, 966-974.	2.5	41
49	New case of interstitial deletion 12(q15â€q21.2) in a girl with facial dysmorphism and mental retardation. American Journal of Medical Genetics, Part A, 2008, 146A, 93-96.	1.2	16
50	Oligosaccharyltransferase-Subunit Mutations in Nonsyndromic Mental Retardation. American Journal of Human Genetics, 2008, 82, 1150-1157.	6.2	130
51	Neurobehavioral Profile and Brain Imaging Study of the 22q13.3 Deletion Syndrome in Childhood. Pediatrics, 2008, 122, e376-e382.	2.1	95
52	Polymorphisms of coding trinucleotide repeats of homeogenes in neurodevelopmental psychiatric disorders. Psychiatric Genetics, 2008, 18, 295-301.	1.1	19
53	Mutations in TCF4, Encoding a Class I Basic Helix-Loop-Helix Transcription Factor, Are Responsible for Pitt-Hopkins Syndrome, a Severe Epileptic Encephalopathy Associated with Autonomic Dysfunction. American Journal of Human Genetics, 2007, 80, 988-993.	6.2	264
54	Incidence and clinical features of X-linked Cornelia de Lange syndrome due toSMC1L1 mutations. Human Mutation, 2007, 28, 205-206.	2.5	71

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55	Tequila, a Neurotrypsin Ortholog, Regulates Long-Term Memory Formation in Drosophila. Science, 2006, 313, 851-853.	12.6	74
56	Interstitial 9q22.3 microdeletion: clinical and molecular characterisation of a newly recognised overgrowth syndrome. European Journal of Human Genetics, 2006, 14, 759-767.	2.8	71
57	Father-to-daughter transmission of Cornelia de Lange syndrome caused by a mutation in the 5â€² untranslated region of theNIPBL Gene. Human Mutation, 2006, 27, 731-735.	2.5	58
58	A rapid microarray based whole genome analysis for detection of uniparental disomy. Human Mutation, 2005, 26, 153-159.	2.5	53
59	Clinical and molecular overlap in overgrowth syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2005, 137C, 4-11.	1.6	39
60	Impaired Mitochondrial Glutamate Transport in Autosomal Recessive Neonatal Myoclonic Epilepsy. American Journal of Human Genetics, 2005, 76, 334-339.	6.2	149
61	Molecular karyotyping in human constitutional cytogenetics. European Journal of Medical Genetics, 2005, 48, 214-231.	1.3	24
62	Paradoxical NSD1 Mutations in Beckwith-Wiedemann Syndrome and 11p15 Anomalies in Sotos Syndrome. American Journal of Human Genetics, 2004, 74, 715-720.	6.2	110
63	Extracellular proteases and their inhibitors ingenetic diseases of the central nervous system. Human Molecular Genetics, 2003, 12, R195-R200.	2.9	33
64	Truncating Neurotrypsin Mutation in Autosomal Recessive Nonsyndromic Mental Retardation. Science, 2002, 298, 1779-1781.	12.6	176
65	Fluorescence Genotyping for Screening Cryptic Telomeric Rearrangements. , 2002, 204, 181-190.		4
66	Partial maternal heterodisomy of chromosome 17q25 in a case of severe mental retardation. Human Genetics, 2001, 108, 511-515.	3.8	9
67	A novel automated strategy for screening cryptic telomeric rearrangements in children with idiopathic mental retardation. European Journal of Human Genetics, 2001, 9, 319-327.	2.8	55
68	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
69	The human EZH2 gene: genomic organisation and revised mapping in 7q35 within the critical region for malignant myeloid disorders. European Journal of Human Genetics, 2000, 8, 174-180.	2.8	86
70	Cloning of Z39Ig, a novel gene with immunoglobulin-like domains located on human chromosome X. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2000, 1492, 522-525.	2.4	38
71	A polymorphic microsatellite XNP-GT in the XNP/ATRX gene's promotor allows familial indirect diagnosis. Human Mutation, 1999, 14, 448-448.	2.5	0
72	Evidence for a new X-linked mental retardation gene in Xp21-Xp22: Clinical and molecular data in one family. , 1999, 83, 132-137.		8

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73	Genetic Basis of Mental Retardation. , 1999, , .		0
74	An imprinted antisense RNA overlaps UBE3A and a second maternally expressed transcript. Nature Genetics, 1998, 19, 15-16.	21.4	292
75	Determination of the Genomic Structure of the XNP/ATRX Gene Encoding a Potential Zinc Finger Helicase. Genomics, 1997, 43, 149-155.	2.9	60
76	Expression and mapping of the mouse S7/Pmsc2 gene, homolog of an essential mitotic gene in yeast. Mammalian Genome, 1997, 8, 352-354.	2.2	0
77	Developmental Changes of the 26 S Proteasome in Abdominal Intersegmental Muscles of Manduca sexta during Programmed Cell Death. Journal of Biological Chemistry, 1995, 270, 1850-1858.	3.4	146
78	Molecular analysis of four males with mental retardation and deletions of Xq21 places the putative MR region in Xq21.1 between DXS233 and CHM. Human Molecular Genetics, 1995, 4, 1465-1466.	2.9	26
79	Construction of a YAC contig spanning the Xq13.3 subband. Genomics, 1995, 26, 115-122.	2.9	18
80	Rapid physical mapping of YAC inserts by random integration of I-Sce I sites. Human Molecular Genetics, 1993, 2, 265-271.	2.9	14
81	Biochemical, genetic and molecular characterization of new respiratory-deficient mutants inChlamydomonas reinhardtii. Plant Molecular Biology, 1992, 18, 759-772.	3.9	68
82	Sequence of a segment of yeast chromosome XI identifies a new mitochondrial carrier,a new member of the G protein family, and a protein with thePAAKK motif of the H1 histones. Yeast, 1992, 8, 325-336.	1.7	22
83	XI. Yeast sequencing reports. The sequence of a 9Â·3 kb segment located on the left arm of the yeast chromosome XI reveals five open reading frames including theCCE1 gene and putative products related to MYO2 and to the ribosomal protein L10. Yeast, 1992, 8, 987-995.	1.7	17
84	The apocytochrome b gene of Chlamydomonas smithii contains a mobile intron related to both Saccharomyces and Neurospora introns. Molecular Genetics and Genomics, 1990, 223, 288-296.	2.4	64
85	Purification and characterization of thein vitroactivity of I-SceI, a novel and highly specific endonuclease encoded by a group I intron. Nucleic Acids Research, 1990, 18, 1407-1413.	14.5	184