Laurent Pasquier

List of Publications by Year
in descending order

103 papers	5,701 citations	61984 43 h-index	85541 71 g-index
108 all docs	108 docs citations	108 times ranked	9687 citing authors

#	Article	IF	Citations
1	How do non-geneticist physicians deal with genetic tests? A qualitative analysis. European Journal of Human Genetics, 2022, 30, 320-331.	2.8	5
2	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics, 2022, 30, 567-576.	2.8	12
3	Informing relatives of their genetic risk: an examination of the Belgian legal context. European Journal of Human Genetics, 2022, 30, 766-771.	2.8	8
4	A systematic review of the views of healthcare professionals on the scope of preimplantation genetic testing. Journal of Community Genetics, 2022, 13, 1-11.	1.2	5
5	Child with a mild CHIME syndrome phenotype and carrying a novel p.(Asp52Asn) PIGL pathogenic variant in association with the previously reported p.(Leu167Pro) variant: A case report. Pediatric Dermatology, 2022, , .	0.9	1
6	<scp>IQSEC2</scp>â€related encephalopathy in males due to missense variants in the pleckstrin homology domain. Clinical Genetics, 2022, 102, 72-77.	2.0	4
7	<scp>Skrabanâ€Deardorff</scp> syndrome: Six new cases of <scp>WDR26</scp>â€related disease and expansion of the clinical phenotype. Clinical Genetics, 2021, 99, 732-739.	2.0	4
8	High rate of hypomorphic variants as the cause of inherited ataxia and related diseases: study of a cohort of 366 families. Genetics in Medicine, 2021, 23, 2160-2170.	2.4	13
9	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
10	<scp>GREB1L</scp> variants in familial and sporadic hereditary urogenital adysplasia and <scp>Mayerâ€Rokitanskyâ€Kusterâ€Hauser</scp> syndrome. Clinical Genetics, 2020, 98, 126-137.	2.0	32
11	Hearing loss in inherited peripheral neuropathies: Molecular diagnosis by NGS in a French series. Molecular Genetics & Genomic Medicine, 2019, 7, e839.	1.2	17
12	Evaluation of the template letter regarding the disclosure of genetic information within the family in France. Journal of Community Genetics, 2019, 10, 489-499.	1.2	7
13	Searching for secondary findings: considering actionability and preserving the right not to know. European Journal of Human Genetics, 2019, 27, 1481-1484.	2.8	13
14	Population genetic screening: current issues in a European country. European Journal of Human Genetics, 2019, 27, 1321-1323.	2.8	2
15	Targeted panel sequencing establishes the implication of planar cell polarity pathway and involves new candidate genes in neural tube defect disorders. Human Genetics, 2019, 138, 363-374.	3.8	15
16	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	3.2	46
17	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
18	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80

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19	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. Brain, 2019, 142, 35-49.	7.6	44
20	Wiedemannâ€Steiner syndrome as a major cause of syndromic intellectual disability: A study of 33 French cases. Clinical Genetics, 2018, 94, 141-152.	2.0	57
21	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
22	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
23	Loss of function IFT27 variants associated with an unclassified lethal fetal ciliopathy with renal agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1610-1613.	1.2	16
24	22q11.2 rearrangements found in women with low ovarian reserve and premature ovarian insufficiency. Journal of Human Genetics, 2018, 63, 691-698.	2.3	4
25	Using medical exome sequencing to identify the causes of neurodevelopmental disorders: Experience of 2 clinical units and 216 patients. Clinical Genetics, 2018, 93, 567-576.	2.0	82
26	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
27	Further refinement of COL4A1 and COL4A2 related cortical malformations. European Journal of Medical Genetics, 2018, 61, 765-772.	1.3	29
28	A de novo variant in ADGRL2 suggests a novel mechanism underlying the previously undescribed association of extreme microcephaly with severely reduced sulcation and rhombencephalosynapsis. Acta Neuropathologica Communications, 2018, 6, 109.	5.2	20
29	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
30	Fetal costello syndrome with neuromuscular spindles excess and p.Gly12Val HRAS mutation. European Journal of Medical Genetics, 2017, 60, 395-398.	1.3	7
31	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
32	Fifteen years of research on oralâ€“facialâ€“digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
33	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
34	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. Acta Neuropathologica, 2017, 134, 889-904.	7.7	42
35	Role of Genetics in the Etiology of Autistic Spectrum Disorder: Towards a Hierarchical Diagnostic Strategy. International Journal of Molecular Sciences, 2017, 18, 618.	4.1	24
36	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. American Journal of Medical Genetics, Part A, 2016, 170, 116-129.	1.2	19

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37	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. Human Mutation, 2016, 37, 1329-1339.	2.5	56
38	A series of 38 novel germline and somatic mutations of <scp>NIPBL</scp> in Cornelia de Lange syndrome. Clinical Genetics, 2016, 89, 584-589.	2.0	20
39	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
40	Karyotype is not dead (yet)!. European Journal of Medical Genetics, 2016, 59, 11-15.	1.3	19
41	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
42	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. Journal of Medical Genetics, 2015, 52, 61-70.	3.2	74
43	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1252-1261.	1.2	20
44	Gene dosage effects in 46, XY DSD: usefulness of CGH technologies for diagnosis. Journal of Assisted Reproduction and Genetics, 2015, 32, 287-291.	2.5	8
45	Search for <scp>ReCQL4</scp> mutations in 39 patients genotyped for suspected Rothmundâ€“Thomson/Ballerâ€Gerold syndromes. Clinical Genetics, 2015, 87, 244-251.	2.0	22
46	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
47	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
48	Extensive investigation of the IGF2/H19 imprinting control region reveals novel OCT4/SOX2 binding site defects associated with specific methylation patterns in Beckwith-Wiedemann syndrome. Human Molecular Genetics, 2014, 23, 5763-5773.	2.9	58
49	Delineation ofEFTUD2Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. Human Mutation, 2014, 35, 478-485.	2.5	50
50	Inversion duplication deletions involving the long arm of chromosome 13: Phenotypic description of additional three fetuses and genotypeâ€“phenotype correlation. American Journal of Medical Genetics, Part A, 2014, 164, 2504-2509.	1.2	13
51	Deep intronic <scp>KRIT1</scp> mutation in a family with clinically silent multiple cerebral cavernous malformations. Clinical Genetics, 2014, 86, 585-588.	2.0	16
52	Split hand/foot malformation with longâ€bone deficiency and <scp>BHLHA9</scp> duplication: report of 13 new families. Clinical Genetics, 2014, 85, 464-469.	2.0	20
53	The oral-facial-digital syndrome gene C2CD3 encodes a positive regulator of centriole elongation. Nature Genetics, 2014, 46, 905-911.	21.4	121
54	CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. Orphanet Journal of Rare Diseases, 2014, 9, 74.	2.7	34

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55	Postnatal diagnosis of 9q interstitial imbalances involving PTCH1, resulting from a familial intrachromosomal insertion. European Journal of Medical Genetics, 2014, 57, 195-199.	1.3	2
56	Molecular and clinical characterization of 25 individuals with exonic deletions of NRXN1 and comprehensive review of the literature. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 388-403.	1.7	93
57	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. European Journal of Medical Genetics, 2013, 56, 301-308.	1.3	17
58	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. Clinical Genetics, 2013, 84, 507-521.	2.0	23
59	Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. European Journal of Human Genetics, 2013, 21, 855-863.	2.8	52
60	Array-CGH Analysis Suggests Genetic Heterogeneity in Rhombencephalosynapsis. Molecular Syndromology, 2013, 4, 267-272.	0.8	19
61	Clinical utility gene card for: Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome. European Journal of Human Genetics, 2012, 20, 1-3.	2.8	20
62	Binder phenotype in mothers affected with autoimmune disorders. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 1413-1418.	1.5	8
63	Molecular characterization of 1q44 microdeletion in 11 patients reveals three candidate genes for intellectual disability and seizures. American Journal of Medical Genetics, Part A, 2012, 158A, 1633-1640.	1.2	63
64	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
65	Utero-vaginal aplasia (Mayer-Rokitansky-KÃ¼ster-Hauser syndrome) associated with deletions in known DiGeorge or DiGeorge-like loci. Orphanet Journal of Rare Diseases, 2011, 6, 9.	2.7	48
66	GLI3 is rarely implicated in OFD syndromes with midline abnormalities. Human Mutation, 2011, 32, 1332-1333.	2.5	5
67	New findings for phenotype-genotype correlations in a large European series of holoprosencephaly cases. Journal of Medical Genetics, 2011, 48, 752-760.	3.2	90
68	Mutation update for theCSB/ERCC6andCSA/ERCC8genes involved in Cockayne syndrome. Human Mutation, 2010, 31, 113-126.	2.5	193
69	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
70	Genetic counseling and â€œmolecularâ€•prenatal diagnosis of holoprosencephaly (HPE). American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 191-196.	1.6	47
71	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
72	C2orf37 mutational spectrum in Woodhouseâ€“Sakati syndrome patients. Clinical Genetics, 2010, 78, 585-590.	2.0	41

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73	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. European Journal of Medical Genetics, 2010, 53, 66-75.	1.3	29
74	2q23.1 microdeletion identified by array comparative genomic hybridisation: an emerging phenotype with Angelman-like features?. Journal of Medical Genetics, 2009, 46, 847-855.	3.2	43
75	Penetrance of marked cognitive impairment in older male carriers of the FMR1 gene premutation. Journal of Medical Genetics, 2009, 46, 818-824.	3.2	61
76	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
77	Array-CGH analysis indicates a high prevalence of genomic rearrangements in holoprosencephaly: An updated map of candidate loci. Human Mutation, 2009, 30, 1175-1182.	2.5	46
78	Rhombencephalosynapsis and related anomalies: a neuropathological study of 40 fetal cases. Acta Neuropathologica, 2009, 117, 185-200.	7.7	96
79	Twelve new patients with 13q deletion syndrome: Genotypeâ€“phenotype analyses in progress. European Journal of Medical Genetics, 2009, 52, 41-46.	1.3	80
80	Cryptic genomic imbalances in de novo and inherited apparently balanced chromosomal rearrangements: Array CGH study of 47 unrelated cases. European Journal of Medical Genetics, 2009, 52, 291-296.	1.3	89
81	Multi-view renderer for auto-stereoscopic mobile devices. , 2009, , .		2
82	Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in TUBA1A. Brain, 2008, 131, 2304-2320.	7.6	98
83	Phenotypic variability of a 4q34â†'qter inherited deletion: MRKH syndrome in the daughter, cardiac defect and Fallopian tube cancer in the mother. European Journal of Medical Genetics, 2007, 50, 66-72.	1.3	34
84	MLPA screening reveals novel subtelomeric rearrangements in holoprosencephaly. Human Mutation, 2007, 28, 1189-1197.	2.5	25
85	Holoprosencephaly. Orphanet Journal of Rare Diseases, 2007, 2, 8.	2.7	299
86	Large spectrum of lissencephaly and pachygyria phenotypes resulting from de novo missense mutations in tubulin alpha 1A (TUBA1A). Human Mutation, 2007, 28, 1055-1064.	2.5	213
87	Spectrum and distribution of MECP2 mutations in 424 Rett syndrome patients: a molecular update. European Journal of Medical Genetics, 2006, 49, 9-18.	1.3	64
88	The Incidence of Rett Syndrome in France. Pediatric Neurology, 2006, 34, 372-375.	2.1	44
89	The Mayer-Rokitansky-KÃ¼ster-Hauser syndrome (congenital absence of uterus and vagina) â€“ phenotypic manifestations and genetic approaches. Journal of Negative Results in BioMedicine, 2006, 5, 1.	1.4	145
90	Molecular evaluation of foetuses with holoprosencephaly shows high incidence of microdeletions in the HPE genes. Human Genetics, 2006, 119, 1-8.	3.8	52

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91	Multicolour FISH and quantitative PCR can detect submicroscopic deletions in holoprosencephaly patients with a normal karyotype. Journal of Medical Genetics, 2006, 43, 496-500.	3.2	40
92	Vertebral anomalies and cartilaginous tracheal sleeve in three patients with Pfeiffer syndrome carrying the S351C FGFR2 mutation. Clinical Genetics, 2005, 68, 179-181.	2.0	28
93	Wide clinical variability among 13 new Cockayne syndrome cases confirmed by biochemical assays. Archives of Disease in Childhood, 2005, 91, 178-182.	1.9	39
94	First occurrence of aprosencephaly/atelencephaly and holoprosencephaly in a family with a SIX3 gene mutation and phenotype/genotype correlation in our series of SIX3 mutations. Journal of Medical Genetics, 2005, 42, e4-e4.	3.2	42
95	Sporadic optic atrophy due to synonymous codon change altering mRNA splicing of OPA1. Clinical Genetics, 2004, 67, 102-103.	2.0	9
96	Congenital diaphragmatic hernia: prenatal diagnosis permits immediate intensive care with high survival rate in isolated cases. A populationâ€based study. Prenatal Diagnosis, 2004, 24, 487-493.	2.3	51
97	Phenotypic and molecular variability of the holoprosencephalic spectrum. American Journal of Medical Genetics, Part A, 2004, 129A, 21-24.	1.2	61
98	Molecular screening ofSHH,ZIC2,SIX3, andTGIF genes in patients with features of holoprosencephaly spectrum: Mutation review and genotype-phenotype correlations. Human Mutation, 2004, 24, 43-51.	2.5	128
99	Molecular screening of the TGIF gene in holoprosencephaly: identification of two novel mutations. Human Genetics, 2003, 112, 131-134.	3.8	43
100	The association of autosomal dominant optic atrophy and moderate deafness may be due to the R445H mutation in the OPA1 gene. American Journal of Ophthalmology, 2003, 136, 1170-1171.	3.3	58
101	Perinatal management and outcome of prenatally diagnosed congenital diaphragmatic hernia: a 1995â€“2000 series in Rennes University Hospital. Prenatal Diagnosis, 2002, 22, 988-994.	2.3	39
102	Overgrowth and trisomy 15q26.1-qter including the IGF1 receptor gene: report of two families and review of the literature. European Journal of Human Genetics, 2002, 10, 699-706.	2.8	44
103	A new mutation in the six-domain of SIX3 gene causes holoprosencephaly. European Journal of Human Genetics, 2000, 8, 797-800.	2.8	55