

# Laurent Pasquier

## List of Publications by Year in descending order

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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	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. <i>Nature</i> , 2011, 478, 97-102.	27.8	394
2	Holoprosencephaly. <i>Orphanet Journal of Rare Diseases</i> , 2007, 2, 8.	2.7	299
3	Large spectrum of lissencephaly and pachygyria phenotypes resulting from de novo missense mutations in tubulin alpha 1A ( <i>TUBA1A</i> ). <i>Human Mutation</i> , 2007, 28, 1055-1064.	2.5	213
4	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. <i>Human Mutation</i> , 2010, 31, E1506-E1518.	2.5	208
5	Mutation update for the <i>CSB</i> / <i>ERCC6</i> and <i>CSA</i> / <i>ERCC8</i> genes involved in Cockayne syndrome. <i>Human Mutation</i> , 2010, 31, 113-126.	2.5	193
6	The Mayer-Rokitansky-Küster-Hauser syndrome (congenital absence of uterus and vagina) – phenotypic manifestations and genetic approaches. <i>Journal of Negative Results in BioMedicine</i> , 2006, 5, 1.	1.4	145
7	De Novo Mutations in Protein Kinase Genes <i>CAMK2A</i> and <i>CAMK2B</i> Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	6.2	136
8	Molecular screening of <i>SHH</i> , <i>ZIC2</i> , <i>SIX3</i> , and <i>TGIF</i> genes in patients with features of holoprosencephaly spectrum: Mutation review and genotype-phenotype correlations. <i>Human Mutation</i> , 2004, 24, 43-51.	2.5	128
9	Mutational, functional, and expression studies of the <i>TCF4</i> gene in Pitt-Hopkins syndrome. <i>Human Mutation</i> , 2009, 30, 669-676.	2.5	126
10	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. <i>Genetics in Medicine</i> , 2016, 18, 49-56.	2.4	125
11	The oral-facial-digital syndrome gene <i>C2CD3</i> encodes a positive regulator of centriole elongation. <i>Nature Genetics</i> , 2014, 46, 905-911.	21.4	121
12	Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in <i>TUBA1A</i> . <i>Brain</i> , 2008, 131, 2304-2320.	7.6	98
13	New insights into genotype-phenotype correlation for <i>GLI3</i> mutations. <i>European Journal of Human Genetics</i> , 2015, 23, 92-102.	2.8	97
14	Rhombencephalosynapsis and related anomalies: a neuropathological study of 40 fetal cases. <i>Acta Neuropathologica</i> , 2009, 117, 185-200.	7.7	96
15	<i>HCN1</i> mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018, 141, 3160-3178.	7.6	96
16	Molecular and clinical characterization of 25 individuals with exonic deletions of <i>NRXN1</i> and comprehensive review of the literature. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2013, 162, 388-403.	1.7	93
17	New findings for phenotype-genotype correlations in a large European series of holoprosencephaly cases. <i>Journal of Medical Genetics</i> , 2011, 48, 752-760.	3.2	90
18	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the <i>TBX-FGF</i> Pathway. <i>American Journal of Human Genetics</i> , 2019, 104, 213-228.	6.2	90

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19	Cryptic genomic imbalances in de novo and inherited apparently balanced chromosomal rearrangements: Array CGH study of 47 unrelated cases. <i>European Journal of Medical Genetics</i> , 2009, 52, 291-296.	1.3	89
20	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	6.2	86
21	Fifteen years of research on oral-facial-digital syndromes: from 1 to 16 causal genes. <i>Journal of Medical Genetics</i> , 2017, 54, 371-380.	3.2	85
22	Using medical exome sequencing to identify the causes of neurodevelopmental disorders: Experience of 2 clinical units and 216 patients. <i>Clinical Genetics</i> , 2018, 93, 567-576.	2.0	82
23	Twelve new patients with 13q deletion syndrome: Genotype-phenotype analyses in progress. <i>European Journal of Medical Genetics</i> , 2009, 52, 41-46.	1.3	80
24	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	2.4	80
25	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. <i>Journal of Medical Genetics</i> , 2015, 52, 61-70.	3.2	74
26	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. <i>Human Genetics</i> , 2017, 136, 463-479.	3.8	66
27	Spectrum and distribution of MECP2 mutations in 424 Rett syndrome patients: a molecular update. <i>European Journal of Medical Genetics</i> , 2006, 49, 9-18.	1.3	64
28	Molecular characterization of 1q44 microdeletion in 11 patients reveals three candidate genes for intellectual disability and seizures. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1633-1640.	1.2	63
29	Phenotypic and molecular variability of the holoprosencephalic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2004, 129A, 21-24.	1.2	61
30	Penetrance of marked cognitive impairment in older male carriers of the FMR1 gene premutation. <i>Journal of Medical Genetics</i> , 2009, 46, 818-824.	3.2	61
31	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. <i>Human Molecular Genetics</i> , 2014, 23, 6069-6080.	2.9	61
32	The association of autosomal dominant optic atrophy and moderate deafness may be due to the R445H mutation in the OPA1 gene. <i>American Journal of Ophthalmology</i> , 2003, 136, 1170-1171.	3.3	58
33	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. <i>Human Molecular Genetics</i> , 2014, 23, 5763-5773.	2.9	58
34	Wiedemann-Steiner syndrome as a major cause of syndromic intellectual disability: A study of 33 French cases. <i>Clinical Genetics</i> , 2018, 94, 141-152.	2.0	57
35	Mutational Spectrum in Holoprosencephaly Shows That FGF is a New Major Signaling Pathway. <i>Human Mutation</i> , 2016, 37, 1329-1339.	2.5	56
36	A new mutation in the six-domain of SIX3 gene causes holoprosencephaly. <i>European Journal of Human Genetics</i> , 2000, 8, 797-800.	2.8	55

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37	Molecular evaluation of foetuses with holoprosencephaly shows high incidence of microdeletions in the HPE genes. <i>Human Genetics</i> , 2006, 119, 1-8.	3.8	52
38	Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. <i>European Journal of Human Genetics</i> , 2013, 21, 855-863.	2.8	52
39	Congenital diaphragmatic hernia: prenatal diagnosis permits immediate intensive care with high survival rate in isolated cases. A population-based study. <i>Prenatal Diagnosis</i> , 2004, 24, 487-493.	2.3	51
40	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	6.2	51
41	Delineation of <i>EFTUD2</i> Haploinsufficiency-Related Phenotypes Through a Series of 36 Patients. <i>Human Mutation</i> , 2014, 35, 478-485.	2.5	50
42	Utero-vaginal aplasia (Mayer-Rokitansky-Küster-Hauser syndrome) associated with deletions in known DiGeorge or DiGeorge-like loci. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 9.	2.7	48
43	Genetic counseling and molecular prenatal diagnosis of holoprosencephaly (HPE). <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2010, 154C, 191-196.	1.6	47
44	Array-CGH analysis indicates a high prevalence of genomic rearrangements in holoprosencephaly: An updated map of candidate loci. <i>Human Mutation</i> , 2009, 30, 1175-1182.	2.5	46
45	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. <i>Journal of Medical Genetics</i> , 2019, 56, 526-535.	3.2	46
46	Further delineation of the <i>MECP2</i> duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. <i>Journal of Medical Genetics</i> , 2018, 55, 359-371.	3.2	45
47	Overgrowth and trisomy 15q26.1-qter including the IGF1 receptor gene: report of two families and review of the literature. <i>European Journal of Human Genetics</i> , 2002, 10, 699-706.	2.8	44
48	The Incidence of Rett Syndrome in France. <i>Pediatric Neurology</i> , 2006, 34, 372-375.	2.1	44
49	Integrated clinical and omics approach to rare diseases: novel genes and oligogenic inheritance in holoprosencephaly. <i>Brain</i> , 2019, 142, 35-49.	7.6	44
50	Molecular screening of the TGIF gene in holoprosencephaly: identification of two novel mutations. <i>Human Genetics</i> , 2003, 112, 131-134.	3.8	43
51	2q23.1 microdeletion identified by array comparative genomic hybridisation: an emerging phenotype with Angelman-like features?. <i>Journal of Medical Genetics</i> , 2009, 46, 847-855.	3.2	43
52	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. <i>Journal of Medical Genetics</i> , 2005, 42, e4-e4.	3.2	42
53	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. <i>Acta Neuropathologica</i> , 2017, 134, 889-904.	7.7	42
54	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor $\beta^2$ Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	1.3	42

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55	<i>C2orf37</i> mutational spectrum in Woodhouseâ€“Sakati syndrome patients. <i>Clinical Genetics</i> , 2010, 78, 585-590.	2.0	41
56	Multicolour FISH and quantitative PCR can detect submicroscopic deletions in holoprosencephaly patients with a normal karyotype. <i>Journal of Medical Genetics</i> , 2006, 43, 496-500.	3.2	40
57	Perinatal management and outcome of prenatally diagnosed congenital diaphragmatic hernia: a 1995â€“2000 series in Rennes University Hospital. <i>Prenatal Diagnosis</i> , 2002, 22, 988-994.	2.3	39
58	Wide clinical variability among 13 new Cockayne syndrome cases confirmed by biochemical assays. <i>Archives of Disease in Childhood</i> , 2005, 91, 178-182.	1.9	39
59	Targeted resequencing identifies <i>PTCH1</i> as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. <i>Genome Research</i> , 2016, 26, 474-485.	5.5	37
60	Phenotypic variability of a 4q34â†’qter inherited deletion: MRKH syndrome in the daughter, cardiac defect and Fallopian tube cancer in the mother. <i>European Journal of Medical Genetics</i> , 2007, 50, 66-72.	1.3	34
61	CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 74.	2.7	34
62	<i>GREB1L</i> variants in familial and sporadic hereditary urogenital adysplasia and Mayerâ€“Rokitanskyâ€“Kusterâ€“Hauser syndrome. <i>Clinical Genetics</i> , 2020, 98, 126-137.	2.0	32
63	Identification of gene copy number variations in patients with mental retardation using array-CGH: Novel syndromes in a large French series. <i>European Journal of Medical Genetics</i> , 2010, 53, 66-75.	1.3	29
64	Further refinement of COL4A1 and COL4A2 related cortical malformations. <i>European Journal of Medical Genetics</i> , 2018, 61, 765-772.	1.3	29
65	Vertebral anomalies and cartilaginous tracheal sleeve in three patients with Pfeiffer syndrome carrying the S351C FGFR2 mutation. <i>Clinical Genetics</i> , 2005, 68, 179-181.	2.0	28
66	MLPA screening reveals novel subtelomeric rearrangements in holoprosencephaly. <i>Human Mutation</i> , 2007, 28, 1189-1197.	2.5	25
67	Role of Genetics in the Etiology of Autistic Spectrum Disorder: Towards a Hierarchical Diagnostic Strategy. <i>International Journal of Molecular Sciences</i> , 2017, 18, 618.	4.1	24
68	Systematic molecular and cytogenetic screening of 100 patients with marfanoid syndromes and intellectual disability. <i>Clinical Genetics</i> , 2013, 84, 507-521.	2.0	23
69	Search for <i>ReCQL4</i> mutations in 39 patients genotyped for suspected Rothmundâ€“Thomson/Ballerâ€“Gerold syndromes. <i>Clinical Genetics</i> , 2015, 87, 244-251.	2.0	22
70	Array-based comparative genomic hybridization identifies a high frequency of copy number variations in patients with syndromic overgrowth. <i>European Journal of Human Genetics</i> , 2010, 18, 227-232.	2.8	20
71	Clinical utility gene card for: Mayerâ€“Rokitanskyâ€“KÅ¼sterâ€“Hauser syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 1-3.	2.8	20
72	Split hand/foot malformation with longâ€“bone deficiency and <i>BHLHA9</i> duplication: report of 13 new families. <i>Clinical Genetics</i> , 2014, 85, 464-469.	2.0	20

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73	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1252-1261.	1.2	20
74	A series of 38 novel germline and somatic mutations of <i>NIPBL</i> in Cornelia de Lange syndrome. <i>Clinical Genetics</i> , 2016, 89, 584-589.	2.0	20
75	A de novo variant in <i>ADGRL2</i> suggests a novel mechanism underlying the previously undescribed association of extreme microcephaly with severely reduced sulcation and rhombencephalosynapsis. <i>Acta Neuropathologica Communications</i> , 2018, 6, 109.	5.2	20
76	Array-CGH Analysis Suggests Genetic Heterogeneity in Rhombencephalosynapsis. <i>Molecular Syndromology</i> , 2013, 4, 267-272.	0.8	19
77	Large national series of patients with Xq28 duplication involving <i>MECP2</i> : Delineation of brain MRI abnormalities in 30 affected patients. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 116-129.	1.2	19
78	Karyotype is not dead (yet)!. <i>European Journal of Medical Genetics</i> , 2016, 59, 11-15.	1.3	19
79	Detailed clinical, genetic and neuroimaging characterization of OFD VI syndrome. <i>European Journal of Medical Genetics</i> , 2013, 56, 301-308.	1.3	17
80	Hearing loss in inherited peripheral neuropathies: Molecular diagnosis by NGS in a French series. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e839.	1.2	17
81	Deep intronic <i>KRIT1</i> mutation in a family with clinically silent multiple cerebral cavernous malformations. <i>Clinical Genetics</i> , 2014, 86, 585-588.	2.0	16
82	Loss of function <i>IFT27</i> variants associated with an unclassified lethal fetal ciliopathy with renal agenesis. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1610-1613.	1.2	16
83	Targeted panel sequencing establishes the implication of planar cell polarity pathway and involves new candidate genes in neural tube defect disorders. <i>Human Genetics</i> , 2019, 138, 363-374.	3.8	15
84	Inversion duplication deletions involving the long arm of chromosome 13: Phenotypic description of additional three fetuses and genotype-phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2504-2509.	1.2	13
85	Searching for secondary findings: considering actionability and preserving the right not to know. <i>European Journal of Human Genetics</i> , 2019, 27, 1481-1484.	2.8	13
86	High rate of hypomorphic variants as the cause of inherited ataxia and related diseases: study of a cohort of 366 families. <i>Genetics in Medicine</i> , 2021, 23, 2160-2170.	2.4	13
87	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. <i>European Journal of Human Genetics</i> , 2022, 30, 567-576.	2.8	12
88	Sporadic optic atrophy due to synonymous codon change altering mRNA splicing of <i>OPA1</i> . <i>Clinical Genetics</i> , 2004, 67, 102-103.	2.0	9
89	Binder phenotype in mothers affected with autoimmune disorders. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2012, 25, 1413-1418.	1.5	8
90	Gene dosage effects in 46, XY DSD: usefulness of CGH technologies for diagnosis. <i>Journal of Assisted Reproduction and Genetics</i> , 2015, 32, 287-291.	2.5	8

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91	Informing relatives of their genetic risk: an examination of the Belgian legal context. <i>European Journal of Human Genetics</i> , 2022, 30, 766-771.	2.8	8
92	Fetal costello syndrome with neuromuscular spindles excess and p.Gly12Val HRAS mutation. <i>European Journal of Medical Genetics</i> , 2017, 60, 395-398.	1.3	7
93	Evaluation of the template letter regarding the disclosure of genetic information within the family in France. <i>Journal of Community Genetics</i> , 2019, 10, 489-499.	1.2	7
94	GLI3 is rarely implicated in OFD syndromes with midline abnormalities. <i>Human Mutation</i> , 2011, 32, 1332-1333.	2.5	5
95	How do non-geneticist physicians deal with genetic tests? A qualitative analysis. <i>European Journal of Human Genetics</i> , 2022, 30, 320-331.	2.8	5
96	A systematic review of the views of healthcare professionals on the scope of preimplantation genetic testing. <i>Journal of Community Genetics</i> , 2022, 13, 1-11.	1.2	5
97	22q11.2 rearrangements found in women with low ovarian reserve and premature ovarian insufficiency. <i>Journal of Human Genetics</i> , 2018, 63, 691-698.	2.3	4
98	<scp>Skrabanâ€Deardorff</scp> syndrome: Six new cases of <scp><i>WDR</i>26</scp>â€related disease and expansion of the clinical phenotype. <i>Clinical Genetics</i> , 2021, 99, 732-739.	2.0	4
99	<scp>IQSEC2</scp>â€related encephalopathy in males due to missense variants in the pleckstrin homology domain. <i>Clinical Genetics</i> , 2022, 102, 72-77.	2.0	4
100	Multi-view renderer for auto-stereoscopic mobile devices. , 2009, , .		2
101	Postnatal diagnosis of 9q interstitial imbalances involving PTCH1, resulting from a familial intrachromosomal insertion. <i>European Journal of Medical Genetics</i> , 2014, 57, 195-199.	1.3	2
102	Population genetic screening: current issues in a European country. <i>European Journal of Human Genetics</i> , 2019, 27, 1321-1323.	2.8	2
103	Child with a mild CHIME syndrome phenotype and carrying a novel p.(Asp52Asn) <i>PIGL</i> pathogenic variant in association with the previously reported p.(Leu167Pro) variant: A case report. <i>Pediatric Dermatology</i> , 2022, , .	0.9	1