Patrick Edery

List of Publications by Year
in descending order

64 papers	6,537 citations	76196 40 h-index	110170 64 g-index
66 all docs	66 docs citations	66 times ranked	9363 citing authors

#	Article	IF	Citations
1	Mutations of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 378-380.	13.7	750
2	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
3	Mutation of the endothelin-3 gene in the Waardenburg-Hirschsprung disease (Shah-Waardenburg) Tj ETQq1 1 0.784314 rgBT /Overlock	9.4	425
4	Quinone-responsive multiple respiratory-chain dysfunction due to widespread coenzyme Q10 deficiency. Lancet, The, 2000, 356, 391-395.	6.3	349
5	Diversity of RET proto-oncogene mutations in familial and sporadic Hirschsprung disease. Human Molecular Genetics, 1995, 4, 1381-1386.	1.4	342
6	New Mutations of CIAS1 That Are Responsible for Muckle-Wells Syndrome and Familial Cold Urticaria: A Novel Mutation Underlies Both Syndromes. American Journal of Human Genetics, 2002, 70, 1498-1506.	2.6	313
7	High cumulative risks of cancer in patients withPTENhamartoma tumour syndrome. Journal of Medical Genetics, 2013, 50, 255-263.	1.5	290
8	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	1.5	229
9	Mutation of the endothelin-receptor B gene in Waardenburg-Hirschsprung disease. Human Molecular Genetics, 1995, 4, 2407-2409.	1.4	214
10	The RET proto-oncogene induces apoptosis: a novel mechanism for Hirschsprung disease. EMBO Journal, 2000, 19, 4056-4063.	3.5	208
11	Association of TALS Developmental Disorder with Defect in Minor Splicing Component U4atac snRNA. Science, 2011, 332, 240-243.	6.0	195
12	Epileptic encephalopathies of the Landauâ€Kleffner and continuous spike and waves during slowâ€wave sleep types: Genomic dissection makes the link with autism. Epilepsia, 2012, 53, 1526-1538.	2.6	148
13	GRIN2A-related disorders: genotype and functional consequence predict phenotype. Brain, 2019, 142, 80-92.	3.7	143
14	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	1.1	125
15	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	1.1	114
16	Infantile Spasms Is Associated with Deletion of the MAGI2 Gene on Chromosome 7q11.23-q21.11. American Journal of Human Genetics, 2008, 83, 106-111.	2.6	108
17	Novel comprehensive diagnostic strategy in Pitt-Hopkins syndrome: Clinical score and further delineation of the TCF4 mutational spectrum. Human Mutation, 2012, 33, 64-72.	1.1	102
18	Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations. Journal of Medical Genetics, 2013, 50, 144-150.	1.5	99

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19	Molecular and in silico analyses of the full-length isoform of usherin identify new pathogenic alleles in Usher type II patients. Human Mutation, 2007, 28, 781-789.	1.1	98
20	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	1.1	90
21	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.	0.7	89
22	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. Journal of Medical Genetics, 2017, 54, 843-851.	1.5	88
23	Functional variants of POC5 identified in patients with idiopathic scoliosis. Journal of Clinical Investigation, 2015, 125, 1124-1128.	3.9	87
24	Ret in human development and oncogenesis. BioEssays, 1997, 19, 389-395.	1.2	74
25	A subset of genomic alterations detected in rolandic epilepsies contains candidate or known epilepsy genes including <scp>GRIN</scp>2A and <scp>PRRT</scp>2. Epilepsia, 2014, 55, 370-378.	2.6	69
26	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	1.1	62
27	Spectrum of epilepsy in terminal 1p36 deletion syndrome. Epilepsia, 2008, 49, 509-515.	2.6	59
28	Molecular characterization of a cohort of 73 patients with infantile spasms syndrome. European Journal of Medical Genetics, 2015, 58, 51-58.	0.7	56
29	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	0.7	56
30	Do estrogens impact adolescent idiopathic scoliosis?. Trends in Endocrinology and Metabolism, 2009, 20, 147-152.	3.1	53
31	Using Genomic Inbreeding Coefficient Estimates for Homozygosity Mapping of Rare Recessive Traits: Application to Taybi-Linder Syndrome. American Journal of Human Genetics, 2006, 79, 62-66.	2.6	48
32	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. European Journal of Human Genetics, 2009, 17, 395-400.	1.4	48
33	Clinical and molecular characterization of 17q21.31 microdeletion syndrome in 14 French patients with mental retardation. European Journal of Medical Genetics, 2011, 54, 144-151.	0.7	48
34	14q12 and severe Rett-like phenotypes: new clinical insights and physical mapping of FOXG1-regulatory elements. European Journal of Human Genetics, 2012, 20, 1216-1223.	1.4	48
35	Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. European Journal of Human Genetics, 2017, 25, 423-431.	1.4	48
36	IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814.	1.5	47

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37	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
38	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.	1.5	46
39	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. European Journal of Medical Genetics, 2010, 53, 303-308.	0.7	44
40	16p13.11 microduplication in 45 new patients: refined clinical significance and genotypeâ€“phenotype correlations. Journal of Medical Genetics, 2020, 57, 301-307.	1.5	44
41	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609.	1.1	43
42	New disease gene location and high genetic heterogeneity in idiopathic scoliosis. European Journal of Human Genetics, 2011, 19, 865-869.	1.4	41
43	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
44	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	1.4	36
45	Long-Term Follow-Up in 12 Children with Pulmonary Arteriovenous Malformations: Confirmation of Hereditary Hemorrhagic Telangiectasia in all Cases. Journal of Pediatrics, 2007, 151, 299-306.	0.9	35
46	Microarray expression profiling identifies genes with altered expression in Adolescent Idiopathic Scoliosis. European Spine Journal, 2013, 22, 1300-1311.	1.0	33
47	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	2.6	32
48	Search for the best indicators for the presence of a VPS13B gene mutation and confirmation of diagnostic criteria in a series of 34 patients genotyped for suspected Cohen syndrome. Journal of Medical Genetics, 2010, 47, 549-553.	1.5	31
49	Novel homozygous missense variant of GRIN1 in two sibs with intellectual disability and autistic features without epilepsy. European Journal of Human Genetics, 2017, 25, 376-380.	1.4	30
50	New insights into minor splicingâ€”a transcriptomic analysis of cells derived from TALS patients. Rna, 2019, 25, 1130-1149.	1.6	27
51	Insulin response dysregulation explains abnormal fat storage and increased risk of diabetes mellitus type 2 in Cohen Syndrome. Human Molecular Genetics, 2015, 24, 6603-6613.	1.4	26
52	TWIST microdeletion identified by array CGH in a patient presenting Saethreâ€“Chotzen phenotype and a complex rearrangement involving chromosomes 2 and 7. European Journal of Medical Genetics, 2008, 51, 156-164.	0.7	21
53	Facial emotion perception by intensity in children and adolescents with 22q11.2 deletion syndrome. European Child and Adolescent Psychiatry, 2016, 25, 297-310.	2.8	20
54	A 7 bp deletion of the RET proto-oncogene in familial Hirschsprungâ€˜s disease. Human Molecular Genetics, 1994, 3, 1439-1440.	1.4	19

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55	Biliary lithiasis in early pregnancy and abnormal development of facial and distal limb bones (Binder) Tj ETQq1 1 0.784314 rgBT /Overlo Molecular Teratology, 2005, 73, 188-193.	1.6	17
56	Xp21 deletion in female patients with intellectual disability: Two new cases and a review of the literature. European Journal of Medical Genetics, 2015, 58, 341-345.	0.7	14
57	The psychological impact of cryptic chromosomal abnormalities diagnosis announcement. European Journal of Medical Genetics, 2013, 56, 585-590.	0.7	13
58	Autoimmune hypoparathyroidism in a 12-year-old girl with McKusick cartilage hair hypoplasia. Pediatric Nephrology, 2009, 24, 2449-2453.	0.9	12
59	Clinical and neuroimaging findings in 33 patients with <scp>MCAP</scp> syndrome: A survey to evaluate relevant endpoints for future clinical trials. Clinical Genetics, 2021, 99, 650-661.	1.0	12
60	Mayer-Rokitansky-KÃ¼nster-Hauser syndrome due to 2q12.1q14.1 deletion: PAX8 the causing gene?. European Journal of Medical Genetics, 2020, 63, 103812.	0.7	11
61	Clinical interpretation of variants identified in RNU4ATAC, a non-coding spliceosomal gene. PLoS ONE, 2020, 15, e0235655.	1.1	8
62	Classifying Ectopia Lentis in Marfan Syndrome into Five Grades of Increasing Severity. Journal of Clinical Medicine, 2020, 9, 721.	1.0	7
63	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	1.1	4
64	Liver cytochrome?c oxidase deficiency in a case of neonatal-onset hepatic failure. European Journal of Pediatrics, 1994, 153, 190-194.	1.3	1