Pierre Bitoun

List of Publications by Year
in descending order

44 papers	3,895 citations	218677 26 h-index	276875 41 g-index
46 all docs	46 docs citations	46 times ranked	5474 citing authors

#	Article	IF	Citations
1	Comprehensive variant spectrum of the CNGA3 gene in patients affected by achromatopsia. Human Mutation, 2022, 43, 832-858.	2.5	8
2	Confirmation of FZD5 implication in a cohort of 50 patients with ocular coloboma. European Journal of Human Genetics, 2021, 29, 131-140.	2.8	10
3	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	3.8	35
4	The supplementary motor area modulates interhemispheric interactions during movement preparation. Human Brain Mapping, 2019, 40, 2125-2142.	3.6	44
5	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	21.4	69
6	Genetic Analysis of â€˜PAX6-Negativeâ€™ Individuals with Aniridia or Gillespie Syndrome. PLoS ONE, 2016, 11, e0153757.	2.5	54
7	Gain-of-Function Mutations inRARBCause Intellectual Disability with Progressive Motor Impairment. Human Mutation, 2016, 37, 786-793.	2.5	34
8	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992.	6.2	81
9	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	4.9	29
10	Congenital mirror movements. Neurology, 2014, 82, 1999-2002.	1.1	52
11	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogryposis Type 5. American Journal of Human Genetics, 2014, 94, 734-744.	6.2	171
12	PIK3R1 Mutations Cause Syndromic Insulin Resistance with Lipoatrophy. American Journal of Human Genetics, 2013, 93, 141-149.	6.2	162
13	Recessive and Dominant Mutations in Retinoic Acid Receptor Beta in Cases with Microphthalmia and Diaphragmatic Hernia. American Journal of Human Genetics, 2013, 93, 765-772.	6.2	86
14	Congenital Macular Dystrophy, Corpus Callosum Agenesis, Hippocampi Hypoplasia â€“ A Novel Neuro-Ophthalmic Syndrome: Case Report. Ophthalmic Genetics, 2012, 33, 39-43.	1.2	4
15	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. European Journal of Human Genetics, 2012, 20, 527-533.	2.8	19
16	Preâ€•and postnatal phenotype of 6p25 deletions involving the FOXC1 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 2430-2438.	1.2	30
17	Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. European Journal of Medical Genetics, 2011, 54, 157-160.	1.3	39
18	Identification of dominant FOXE3 and PAX6 mutations in patients with congenital cataract and aniridia. Molecular Vision, 2010, 16, 1705-11.	1.1	43

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19	BCOR analysis in patients with OFCD and Lenz microphthalmia syndromes, mental retardation with ocular anomalies, and cardiac laterality defects. European Journal of Human Genetics, 2009, 17, 1325-1335.	2.8	85
20	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	6.2	81
21	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. American Journal of Human Genetics, 2007, 80, 550-560.	6.2	316
22	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 632 Td (Report of eight cases including a living child and further evidence for autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 1268-1281.	1.2	43
23	Conseil gÃ©nÃ©tique et mÃ©decine prÃ©dictive. EMC - TraitÃ© De MÃ©decine AKOS, 2006, 1, 1-8.	0.0	0
24	SLC26A4 gene is frequently involved in nonsyndromic hearing impairment with enlarged vestibular aqueduct in Caucasian populations. European Journal of Human Genetics, 2006, 14, 773-779.	2.8	204
25	Blepharophimosis-mental retardation (BMR) syndromes: A proposed clinical classification of the so-called Ohdo syndrome, and delineation of two new BMR syndromes, one X-linked and one autosomal recessive. American Journal of Medical Genetics, Part A, 2006, 140A, 1285-1296.	1.2	73
26	CHD7 gene and non-syndromic cleft lip and palate. American Journal of Medical Genetics, Part A, 2006, 140A, 2110-2114.	1.2	28
27	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. European Journal of Human Genetics, 2005, 13, 302-308.	2.8	216
28	Testing for triallelism: analysis of six BBS genes in a Bardetâ€“Biedl syndrome family cohort. European Journal of Human Genetics, 2005, 13, 607-616.	2.8	69
29	Three patients with hallucal polydactyly and WAGR syndrome, including discordant expression of Wilms tumor in MZ twins. , 2005, 134A, 422-425.		15
30	MÃ¶bius sequence, Robin complex, and hypotonia: Severe expression of brainstem disruption spectrum versus Carey-Fineman-Ziter syndrome. , 2004, 127A, 277-287.		12
31	AICA-Ribosiduria: A Novel, Neurologically Devastating Inborn Error of Purine Biosynthesis Caused by Mutation of ATIC. American Journal of Human Genetics, 2004, 74, 1276-1281.	6.2	138
32	ERGs in female carriers of incomplete congenital stationary night blindness (I-CSNB). A family report. Documenta Ophthalmologica, 2003, 107, 203-212.	2.2	9
33	Refinement of the NHS locus on chromosome Xp22.13 and analysis of five candidate genes. European Journal of Human Genetics, 2002, 10, 516-520.	2.8	23
34	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. American Journal of Human Genetics, 2001, 69, 722-737.	6.2	294
35	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	3.8	31
36	A novel homeobox gene PITX3 is mutated in families with autosomal-dominant cataracts and ASMD. Nature Genetics, 1998, 19, 167-170.	21.4	371

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37	Cloning and characterization of a novel bicoid-related homeobox transcription factor gene, RIEG, involved in Rieger syndrome. Nature Genetics, 1996, 14, 392-399.	21.4	852
38	About the French Language Breastfeeding Literature. Journal of Tropical Pediatrics, 1996, 42, 183-184.	1.5	0
39	Glaucoma with a Larsen-like syndrome. Ophthalmic Genetics, 1994, 15, 133-140.	1.2	6
40	Genetic Heterogeneity of Usher Syndrome Type 1 in French Families. Genomics, 1994, 21, 138-143.	2.9	42
41	Wolfram syndrome: A Report of Four Cases and Review of the Literature. Ophthalmic Genetics, 1994, 15, 77-85.	1.2	7
42	A New Look at the Management of the Oculo-Mandibulo- Facial Syndrome. Ophthalmic Paediatrics and Genetics, 1992, 13, 19-26.	0.4	3
43	A hereditary syndrome with retinopathy and ataxia or deafness in two consanguineous brothers. Ophthalmic Paediatrics and Genetics, 1991, 12, 149-152.	0.4	4
44	A hereditary syndrome association of oculocutaneous albinism, dysmorphic features and short stature. Ophthalmic Paediatrics and Genetics, 1990, 11, 209-213.	0.4	2