

# Pierre Bitoun

## List of Publications by Year in descending order

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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	Cloning and characterization of a novel bicoid-related homeobox transcription factor gene, RIEG, involved in Rieger syndrome. <i>Nature Genetics</i> , 1996, 14, 392-399.	21.4	852
2	A novel homeobox gene PITX3 is mutated in families with autosomal-dominant cataracts and ASMD. <i>Nature Genetics</i> , 1998, 19, 167-170.	21.4	371
3	Mutations in STRA6 Cause a Broad Spectrum of Malformations Including Anophthalmia, Congenital Heart Defects, Diaphragmatic Hernia, Alveolar Capillary Dysplasia, Lung Hypoplasia, and Mental Retardation. <i>American Journal of Human Genetics</i> , 2007, 80, 550-560.	6.2	316
4	CNGA3 Mutations in Hereditary Cone Photoreceptor Disorders. <i>American Journal of Human Genetics</i> , 2001, 69, 722-737.	6.2	294
5	CNGB3 mutations account for 50% of all cases with autosomal recessive achromatopsia. <i>European Journal of Human Genetics</i> , 2005, 13, 302-308.	2.8	216
6	SLC26A4 gene is frequently involved in nonsyndromic hearing impairment with enlarged vestibular aqueduct in Caucasian populations. <i>European Journal of Human Genetics</i> , 2006, 14, 773-779.	2.8	204
7	Mutations in PIEZO2 Cause Gordon Syndrome, Marden-Walker Syndrome, and Distal Arthrogyriposis Type 5. <i>American Journal of Human Genetics</i> , 2014, 94, 734-744.	6.2	171
8	PIK3R1 Mutations Cause Syndromic Insulin Resistance with Lipoatrophy. <i>American Journal of Human Genetics</i> , 2013, 93, 141-149.	6.2	162
9	AICA-Ribosiduria: A Novel, Neurologically Devastating Inborn Error of Purine Biosynthesis Caused by Mutation of ATIC. <i>American Journal of Human Genetics</i> , 2004, 74, 1276-1281.	6.2	138
10	Recessive and Dominant Mutations in Retinoic Acid Receptor Beta in Cases with Microphthalmia and Diaphragmatic Hernia. <i>American Journal of Human Genetics</i> , 2013, 93, 765-772.	6.2	86
11	BCOR analysis in patients with OFCD and Lenz microphthalmia syndromes, mental retardation with ocular anomalies, and cardiac laterality defects. <i>European Journal of Human Genetics</i> , 2009, 17, 1325-1335.	2.8	85
12	Identification of CANT1 Mutations in Desbuquois Dysplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 706-710.	6.2	81
13	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. <i>American Journal of Human Genetics</i> , 2016, 98, 981-992.	6.2	81
14	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. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1285-1296.	1.2	73
15	Testing for triallelism: analysis of six BBS genes in a Bardet-Biedl syndrome family cohort. <i>European Journal of Human Genetics</i> , 2005, 13, 607-616.	2.8	69
16	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. <i>Nature Genetics</i> , 2017, 49, 511-514.	21.4	69
17	Genetic Analysis of "PAX6-Negative"™ Individuals with Aniridia or Gillespie Syndrome. <i>PLoS ONE</i> , 2016, 11, e0153757.	2.5	54
18	Congenital mirror movements. <i>Neurology</i> , 2014, 82, 1999-2002.	1.1	52

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19	The supplementary motor area modulates interhemispheric interactions during movement preparation. <i>Human Brain Mapping</i> , 2019, 40, 2125-2142.	3.6	44
20	The PDAC syndrome (pulmonary hypoplasia/agenesis, diaphragmatic hernia/eventration,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 712 Td ( Report of eight cases including a living child and further evidence for autosomal recessive inheritance. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1268-1281.	1.2	43
21	Identification of dominant FOXE3 and PAX6 mutations in patients with congenital cataract and aniridia. <i>Molecular Vision</i> , 2010, 16, 1705-11.	1.1	43
22	Genetic Heterogeneity of Usher Syndrome Type 1 in French Families. <i>Genomics</i> , 1994, 21, 138-143.	2.9	42
23	Molecular diagnosis reveals genetic heterogeneity for the overlapping MKKS and BBS phenotypes. <i>European Journal of Medical Genetics</i> , 2011, 54, 157-160.	1.3	39
24	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. <i>Human Genetics</i> , 2019, 138, 1051-1069.	3.8	35
25	Gain-of-Function Mutations in <i>RARB</i> Cause Intellectual Disability with Progressive Motor Impairment. <i>Human Mutation</i> , 2016, 37, 786-793.	2.5	34
26	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. <i>Human Genetics</i> , 2001, 108, 51-54.	3.8	31
27	Pre- and postnatal phenotype of 6p25 deletions involving the <i>FOXC1</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2430-2438.	1.2	30
28	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. <i>Molecular Autism</i> , 2015, 6, 19.	4.9	29
29	CHD7 gene and non-syndromic cleft lip and palate. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 2110-2114.	1.2	28
30	Refinement of the NHS locus on chromosome Xp22.13 and analysis of five candidate genes. <i>European Journal of Human Genetics</i> , 2002, 10, 516-520.	2.8	23
31	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. <i>European Journal of Human Genetics</i> , 2012, 20, 527-533.	2.8	19
32	Three patients with hallucal polydactyly and WAGR syndrome, including discordant expression of Wilms tumor in MZ twins. , 2005, 134A, 422-425.		15
33	MÃ¸bius sequence, Robin complex, and hypotonia: Severe expression of brainstem disruption spectrum versus Carey-Fineman-Ziter syndrome. , 2004, 127A, 277-287.		12
34	Confirmation of FZD5 implication in a cohort of 50 patients with ocular coloboma. <i>European Journal of Human Genetics</i> , 2021, 29, 131-140.	2.8	10
35	ERGs in female carriers of incomplete congenital stationary night blindness (I-CSNB). A family report. <i>Documenta Ophthalmologica</i> , 2003, 107, 203-212.	2.2	9
36	Comprehensive variant spectrum of the <i>CNGA3</i> gene in patients affected by achromatopsia. <i>Human Mutation</i> , 2022, 43, 832-858.	2.5	8

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37	Wolfram syndrome: A Report of Four Cases and Review of the Literature. <i>Ophthalmic Genetics</i> , 1994, 15, 77-85.	1.2	7
38	Glaucoma with a Larsen-like syndrome. <i>Ophthalmic Genetics</i> , 1994, 15, 133-140.	1.2	6
39	A hereditary syndrome with retinopathy and ataxia or deafness in two consanguineous brothers. <i>Ophthalmic Paediatrics and Genetics</i> , 1991, 12, 149-152.	0.4	4
40	Congenital Macular Dystrophy, Corpus Callosum Agenesis, Hippocampi Hypoplasia – A Novel Neuro-Ophthalmic Syndrome: Case Report. <i>Ophthalmic Genetics</i> , 2012, 33, 39-43.	1.2	4
41	A New Look at the Management of the Oculo-Mandibulo- Facial Syndrome. <i>Ophthalmic Paediatrics and Genetics</i> , 1992, 13, 19-26.	0.4	3
42	A hereditary syndrome association of oculocutaneous albinism, dysmorphic features and short stature. <i>Ophthalmic Paediatrics and Genetics</i> , 1990, 11, 209-213.	0.4	2
43	About the French Language Breastfeeding Literature. <i>Journal of Tropical Pediatrics</i> , 1996, 42, 183-184.	1.5	0
44	Conseil génétique et médecine prédictive. <i>EMC - Traité De Médecine AKOS</i> , 2006, 1, 1-8.	0.0	0