Kinga M Bujakowska

List of Publications by Year
in descending order

43 papers	2,187 citations	304701 22 h-index	361001 35 g-index
50 all docs	50 docs citations	50 times ranked	3054 citing authors

#	Article	IF	Citations
1	The importance of automation in genetic diagnosis: Lessons from analyzing an inherited retinal degeneration cohort with the Mendelian Analysis Toolkit (MATK). Genetics in Medicine, 2022, 24, 332-343.	2.4	7
2	Novel RCBTB1 variants causing later-onset non-syndromic retinal dystrophy with macular chorioretinal atrophy. Ophthalmic Genetics, 2022, , 1-8.	1.2	2
3	Fundamentals of Genetics. , 2022, , 2815-2846.		0
4	<scp>WDR34</scp>, a candidate gene for nonâ€syndromic rodâ€cone dystrophy. Clinical Genetics, 2021, 99, 298-302.	2.0	7
5	Broadening INPP5E phenotypic spectrum: detection of rare variants in syndromic and non-syndromic IRD. Npj Genomic Medicine, 2021, 6, 53.	3.8	8
6	A Hidden Structural Variation in a Known IRD Gene: A Cautionary Tale of Two New Disease Candidate Genes. Journal of Physical Education and Sports Management, 2021, , mcs.a006131.	1.2	0
7	Genetic testing of various eye disorders. , 2020, , 239-258.		0
8	Moving Towards PDE6A Gene Supplementation Therapy. JAMA Ophthalmology, 2020, 138, 1251.	2.5	0
9	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
10	Expanding the phenotypic spectrum in RDH12-associated retinal disease. Journal of Physical Education and Sports Management, 2020, 6, a004754.	1.2	16
11	Copy-number variation contributes 9% of pathogenicity in the inherited retinal degenerations. Genetics in Medicine, 2020, 22, 1079-1087.	2.4	59
12	A combined RNA-seq and whole genome sequencing approach for identification of non-coding pathogenic variants in single families. Human Molecular Genetics, 2020, 29, 967-979.	2.9	12
13	Contribution of noncoding pathogenic variants to RPGRIP1-mediated inherited retinal degeneration. Genetics in Medicine, 2019, 21, 694-704.	2.4	27
14	Longitudinal Clinical Follow-up and Genetic Spectrum of Patients With Rod-Cone Dystrophy Associated With Mutations inPDE6AandPDE6B. JAMA Ophthalmology, 2019, 137, 669.	2.5	32
15	De novo variants in HK1 associated with neurodevelopmental abnormalities and visual impairment. European Journal of Human Genetics, 2019, 27, 1081-1089.	2.8	19
16	Changes in extracellular matrix cause RPE cells to make basal deposits and activate the alternative complement pathway. Human Molecular Genetics, 2018, 27, 147-159.	2.9	58
17	Ift172 conditional knock-out mice exhibit rapid retinal degeneration and protein trafficking defects. Human Molecular Genetics, 2018, 27, 2012-2024.	2.9	21
18	Clinical Features of a Retinopathy Associated With a Dominant Allele of the RGR Gene. , 2018, 59, 4812.		9

#	Article	IF	Citations
19	Special Issue Introduction: Inherited Retinal Disease: Novel Candidate Genes, Genotypeâ€“Phenotype Correlations, and Inheritance Models. Genes, 2018, 9, 215.	2.4	58
20	Photoreceptor Cilia and Retinal Ciliopathies. Cold Spring Harbor Perspectives in Biology, 2017, 9, a028274.	5.5	154
21	Copy-number variation is an important contributor to the genetic causality of inherited retinal degenerations. Genetics in Medicine, 2017, 19, 643-651.	2.4	51
22	The importance of genetic testing as demonstrated by two cases of -associated retinal generation misdiagnosed as LCA. Molecular Vision, 2017, 23, 695-706.	1.1	13
23	Reevaluation of the Retinal Dystrophy Due to Recessive Alleles of RGR With the Discovery of a Cis-Acting Mutation in CDHR1. , 2016, 57, 4806.		25
24	Efficient In Silico Identification of a Common Insertion in the MAK Gene which Causes Retinitis Pigmentosa. PLoS ONE, 2015, 10, e0142614.	2.5	14
25	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
26	Panel-based genetic diagnostic testing for inherited eye diseases is highly accurate and reproducible, and more sensitive for variant detection, than exome sequencing. Genetics in Medicine, 2015, 17, 253-261.	2.4	216
27	Targeted Exon Sequencing in Usher Syndrome Type I. Investigative Ophthalmology and Visual Science, 2014, 55, 8488-8496.	3.3	24
28	The familial dementia gene revisited: a missense mutation revealed by whole-exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. Human Molecular Genetics, 2014, 23, 491-501.	2.9	29
29	Mutations in Pre-mRNA Processing Factors 3, 8, and 31 Cause Dysfunction of the Retinal Pigment Epithelium. American Journal of Pathology, 2014, 184, 2641-2652.	3.8	62
30	Lrit3 Deficient Mouse (nob6): A Novel Model of Complete Congenital Stationary Night Blindness (cCSNB). PLoS ONE, 2014, 9, e90342.	2.5	50
31	Whole-Exome Sequencing Identifies LRIT3 Mutations as a Cause of Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2013, 92, 67-75.	6.2	120
32	Further Insights Into GPR179: Expression, Localization, and Associated Pathogenic Mechanisms Leading to Complete Congenital Stationary Night Blindness. , 2013, 54, 8041.		20
33	A Study into the Evolutionary Divergence of the Core Promoter Elements of PRPF31 and TFPT. Journal of Molecular and Genetic Medicine: an International Journal of Biomedical Research, 2013, 07, .	0.1	1
34	Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. Orphanet Journal of Rare Diseases, 2012, 7, 8.	2.7	144
35	Whole-Exome Sequencing Identifies Mutations in GPR179 Leading to Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2012, 90, 321-330.	6.2	121
36	CRB1 mutations in inherited retinal dystrophies. Human Mutation, 2012, 33, 306-315.	2.5	153

#	ARTICLE	IF	CITATIONS
37	Three Gene-Targeted Mouse Models of RNA Splicing Factor RP Show Late-Onset RPE and Retinal Degeneration. , 2011, 52, 190.		70
38	A novel DFNB31 mutation associated with Usher type 2 syndrome showing variable degrees of auditory loss in a consanguineous Portuguese family. Molecular Vision, 2011, 17, 1598-606.	1.1	17
39	Prevalence and novelty of PRPF31 mutations in French autosomal dominant rod-cone dystrophy patients and a review of published reports. BMC Medical Genetics, 2010, 11, 145.	2.1	49
40	TRPM1 Is Mutated in Patients with Autosomal-Recessive Complete Congenital Stationary Night Blindness. American Journal of Human Genetics, 2009, 85, 720-729.	6.2	207
41	The CXC-Chemokine CXCL4 Interacts with Integrins Implicated in Angiogenesis. PLoS ONE, 2008, 3, e2657.	2.5	39
42	Disease mechanism for retinitis pigmentosa (RP11) caused by missense mutations in the splicing factor gene PRPF31. Molecular Vision, 2008, 14, 683-90.	1.1	26
43	Mutations in TOPORS Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular Retinal Pigment Epithelium Atrophy. American Journal of Human Genetics, 2007, 81, 1098-1103.	6.2	77