

# Wolfgang Berger

## List of Publications by Year in descending order

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Version: 2024-02-01

41  
papers

1,889  
citations

567281

15  
h-index

330143

37  
g-index

48  
all docs

48  
docs citations

48  
times ranked

2788  
citing authors

#	ARTICLE	IF	CITATIONS
1	The molecular basis of human retinal and vitreoretinal diseases. <i>Progress in Retinal and Eye Research</i> , 2010, 29, 335-375.	15.5	485
2	Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. <i>European Journal of Human Genetics</i> , 2014, 22, 99-104.	2.8	229
3	Vascular Defects and Sensorineural Deafness in a Mouse Model of Norrie Disease. <i>Journal of Neuroscience</i> , 2002, 22, 4286-4292.	3.6	136
4	Role of the Norrie Disease Pseudoglioma Gene in Sprouting Angiogenesis during Development of the Retinal Vasculature. , 2005, 46, 3372.		129
5	Mutations in the candidate gene for Norrie disease. <i>Human Molecular Genetics</i> , 1992, 1, 461-465.	2.9	126
6	Post-mortem whole-exome analysis in a large sudden infant death syndrome cohort with a focus on cardiovascular and metabolic genetic diseases. <i>European Journal of Human Genetics</i> , 2017, 25, 404-409.	2.8	98
7	Ectopic Norrin Induces Growth of Ocular Capillaries and Restores Normal Retinal Angiogenesis in Norrie Disease Mutant Mice. <i>Journal of Neuroscience</i> , 2005, 25, 1701-1710.	3.6	88
8	Mutations in <i>EXOSC2</i> are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. <i>Journal of Medical Genetics</i> , 2016, 53, 419-425.	3.2	69
9	Next generation sequencing based identification of disease-associated mutations in Swiss patients with retinal dystrophies. <i>Scientific Reports</i> , 2016, 6, 28755.	3.3	62
10	Integrin-linked kinase controls retinal angiogenesis and is linked to Wnt signaling and exudative vitreoretinopathy. <i>Nature Communications</i> , 2019, 10, 5243.	12.8	54
11	Exome analysis in 34 sudden unexplained death (SUD) victims mainly identified variants in channelopathy-associated genes. <i>International Journal of Legal Medicine</i> , 2018, 132, 1057-1065.	2.2	38
12	Norrin stimulates cell proliferation in the superficial retinal vascular plexus and is pivotal for the recruitment of mural cells. <i>Human Molecular Genetics</i> , 2012, 21, 2619-2630.	2.9	36
13	Biallelic Mutations in <i>CRB1</i> Underlie Autosomal Recessive Familial Foveal Retinoschisis. , 2016, 57, 2637.		34
14	Arrhythmogenic right ventricular cardiomyopathy: implications of next-generation sequencing in appropriate diagnosis. <i>Europace</i> , 2017, 19, euw098.	1.7	31
15	Post-mortem whole-exome sequencing (WES) with a focus on cardiac disease-associated genes in five young sudden unexplained death (SUD) cases. <i>International Journal of Legal Medicine</i> , 2016, 130, 1011-1021.	2.2	26
16	Identification of Novel and Recurrent Disease-Causing Mutations in Retinal Dystrophies Using Whole Exome Sequencing (WES): Benefits and Limitations. <i>PLoS ONE</i> , 2016, 11, e0158692.	2.5	20
17	Genetic Analysis in a Swiss Cohort of Bilateral Congenital Cataract. <i>JAMA Ophthalmology</i> , 2021, 139, 691.	2.5	18
18	Effects of COMT genotype and tolcapone on lapses of sustained attention after sleep deprivation in healthy young men. <i>Neuropsychopharmacology</i> , 2018, 43, 1599-1607.	5.4	17

#	ARTICLE	IF	CITATIONS
19	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. <i>Genes</i> , 2021, 12, 65.	2.4	16
20	Homozygosity Mapping and Whole Exome Sequencing Reveal a Novel Homozygous COL18A1 Mutation Causing Knobloch Syndrome. <i>PLoS ONE</i> , 2014, 9, e112747.	2.5	15
21	Exome Sequencing in a Swiss Childhood Glaucoma Cohort Reveals <i>CYP1B1</i> and <i>FOXC1</i> Variants as Most Frequent Causes. <i>Translational Vision Science and Technology</i> , 2020, 9, 47.	2.2	15
22	<i>C2orf71</i> Mutations as a Frequent Cause of Autosomal-Recessive Retinitis Pigmentosa: Clinical Analysis and Presentation of 8 Novel Mutations. , 2017, 58, 3840.		13
23	Genotype-phenotype spectrum in isolated and syndromic nanophthalmos. <i>Acta Ophthalmologica</i> , 2021, 99, e594-e607.	1.1	13
24	Impact of Genetic Variant Reassessment on the Diagnosis of Arrhythmogenic Right Ventricular Cardiomyopathy Based on the 2010 Task Force Criteria. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003047.	3.6	13
25	A case-control field study on the relationships among type 2 diabetes, sleepiness and habitual caffeine intake. <i>Journal of Psychopharmacology</i> , 2017, 31, 233-242.	4.0	11
26	Atonal homolog 7 (ATOH7) loss-of-function mutations in predominant bilateral optic nerve hypoplasia. <i>Human Molecular Genetics</i> , 2019, 29, 132-148.	2.9	11
27	Application of WES towards Molecular Investigation of Congenital Cataracts: Identification of Novel Alleles and Genes in a Hospital-Based Cohort of South India. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9569.	4.1	10
28	De Novo Assembly-Based Analysis of RPGR Exon ORF15 in an Indigenous African Cohort Overcomes Limitations of a Standard Next-Generation Sequencing (NGS) Data Analysis Pipeline. <i>Genes</i> , 2020, 11, 800.	2.4	9
29	Long-term consequences of developmental vascular defects on retinal vessel homeostasis and function in a mouse model of Norrie disease. <i>PLoS ONE</i> , 2017, 12, e0178753.	2.5	9
30	Long-Range PCR-Based NGS Applications to Diagnose Mendelian Retinal Diseases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1508.	4.1	8
31	Regulation of ABCA1 by AMD-Associated Genetic Variants and Hypoxia in iPSC-RPE. <i>International Journal of Molecular Sciences</i> , 2022, 23, 3194.	4.1	8
32	Genotype-Phenotype Analysis of a Novel Recessive and a Recurrent Dominant <i>SNRNP200</i> Variant Causing Retinitis Pigmentosa. , 2019, 60, 2822.		7
33	Lack of Paraoxonase 1 Alters Phospholipid Composition, but Not Morphology and Function of the Mouse Retina. , 2014, 55, 4714.		6
34	The timing of auditory sensory deficits in Norrie disease has implications for therapeutic intervention. <i>JCI Insight</i> , 2022, 7, .	5.0	6
35	Cystoid edema, neovascularization and inflammatory processes in the murine Norrin-deficient retina. <i>Scientific Reports</i> , 2018, 8, 5970.	3.3	4
36	Absence of Goniodysgenesis in Patients with Chromosome 13Q Microdeletion-Related Microcoria. <i>Ophthalmology Glaucoma</i> , 2018, 1, 145-147.	1.9	3

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37	Functional Characterization of an In-Frame Deletion in the Basic Domain of the Retinal Transcription Factor ATOH7. <i>International Journal of Molecular Sciences</i> , 2022, 23, 1053.	4.1	2
38	Benefits and outcomes of a new multidisciplinary approach for the management and financing of sudden unexplained death cases in a forensic setting in Switzerland. <i>Forensic Science International</i> , 2022, 334, 111240.	2.2	2
39	Homozygosity for a Novel DOCK7 Variant Due to Segmental Uniparental Isodisomy of Chromosome 1 Associated with Early Infantile Epileptic Encephalopathy (EIEE) and Cortical Visual Impairment. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7382.	4.1	2
40	A new and a reclassified ICF patient without mutations in DNMT3B and its interacting proteins SUMO-1 and UBC9 ( <i>Am J Med Genet</i> 136A: 31-37, 2005). <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 195-195.	1.2	1
41	Unusual retinopathy in a child with severe combined immune deficiency. <i>Ophthalmic Genetics</i> , 2018, 39, 92-94.	1.2	1