Bernhard H F Weber

List of Publications by Year
in descending order

76	4,886	147566 31	114278 63
papers	citations	h-index	g-index

76	76	76	7853
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	9.4	1,167
2	A Deep Learning Algorithm for Prediction of Age-Related Eye Disease Study Severity Scale for Age-Related Macular Degeneration from Color Fundus Photography. Ophthalmology, 2018, 125, 1410-1420.	2.5	365
3	Complement C3 Inhibitor Pegcetacoplan for Geographic Atrophy Secondary to Age-Related Macular Degeneration. Ophthalmology, 2020, 127, 186-195.	2.5	306
4	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	1.1	224
5	Prevalence ofBRCA1/2germline mutations in 21â€...401 families with breast and ovarian cancer. Journal of Medical Genetics, 2016, 53, 465-471.	1.5	179
6	Variation of the group 5 grass pollen allergen content of airborne pollen in relation to geographic location and time in season. Journal of Allergy and Clinical Immunology, 2015, 136, 87-95.e6.	1.5	155
7	Release of Bet v 1 from birch pollen from 5 European countries. Results from the HIALINE study. Atmospheric Environment, 2012, 55, 496-505.	1.9	141
8	Gene panel testing of 5589 <scp>BRCA</scp>1/2â€negative index patients with breast cancer in a routine diagnostic setting: results of the German Consortium for Hereditary Breast and Ovarian Cancer. Cancer Medicine, 2018, 7, 1349-1358.	1.3	126
9	Mutation Spectrum of the ABCA4 Gene in 335 Stargardt Disease Patients From a Multicenter German Cohortâ€”Impact of Selected Deep Intronic Variants and Common SNPs. , 2017, 58, 394.		104
10	Resolving the dark matter of ABCA4 for 1054 Stargardt disease probands through integrated genomics and transcriptomics. Genetics in Medicine, 2020, 22, 1235-1246.	1.1	92
11	Prevalence of pathogenic BRCA1/2 germline mutations among 802 women with unilateral triple-negative breast cancer without family cancer history. BMC Cancer, 2018, 18, 265.	1.1	84
12	Cell-Type-Specific Complement Expression in the Healthy and Diseased Retina. Cell Reports, 2019, 29, 2835-2848.e4.	2.9	81
13	Modelling the Genetic Risk in Age-Related Macular Degeneration. PLoS ONE, 2012, 7, e37979.	1.1	79
14	An Eye on Age-Related Macular Degeneration: The Role of MicroRNAs in Disease Pathology. Molecular Diagnosis and Therapy, 2017, 21, 31-43.	1.6	78
15	Mutations inEXOSC2are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Journal of Medical Genetics, 2016, 53, 419-425.	1.5	69
16	Recombinant Haplotypes Narrow the ARMS2/HTRA1 Association Signal for Age-Related Macular Degeneration. Genetics, 2017, 205, 919-924.	1.2	65
17	A Circulating MicroRNA Profile Is Associated with Late-Stage Neovascular Age-Related Macular Degeneration. PLoS ONE, 2014, 9, e107461.	1.1	62
18	Clinical and Genetic Factors Associated with Progression of Geographic Atrophy Lesions in Age-Related Macular Degeneration. PLoS ONE, 2015, 10, e0126636.	1.1	61

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19	Reticular Pseudodrusen in Sorsby FundusÂDystrophy. Ophthalmology, 2015, 122, 1555-1562.	2.5	58
20	Progression of Late-Onset Stargardt Disease. , 2016, 57, 5186.		57
21	Genome-wide association meta-analysis for early age-related macular degeneration highlights novel loci and insights for advanced disease. BMC Medical Genomics, 2020, 13, 120.	0.7	56
22	Sorsby Fundus Dystrophy: Novel Mutations, Novel Phenotypic Characteristics, and Treatment Outcomes. , 2015, 56, 2664.		55
23	Genetic pleiotropy between age-related macular degeneration and 16 complex diseases and traits. Genome Medicine, 2017, 9, 29.	3.6	52
24	A mega-analysis of expression quantitative trait loci (eQTL) provides insight into the regulatory architecture of gene expression variation in liver. Scientific Reports, 2018, 8, 5865.	1.6	52
25	Migraine and Vasospasm in Glaucoma: Age-Related Evaluation of 2027 Patients With Glaucoma or Ocular Hypertension. , 2015, 56, 7999.		51
26	Y chromosome mosaicism is associated with age-related macular degeneration. European Journal of Human Genetics, 2019, 27, 36-41.	1.4	49
27	The Role of the Complement System in Age-Related Macular Degeneration. Deutsches Ärzteblatt International, 2014, 111, 133-8.	0.6	47
28	A Novel Antibody against Human Properdin Inhibits the Alternative Complement System and Specifically Detects Properdin from Blood Samples. PLoS ONE, 2014, 9, e96371.	1.1	44
29	Recurrent structural variation, clustered sites of selection, and disease risk for the complement factor H (CFH) gene family. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E4433-E4442.	3.3	43
30	Synonymous variants in HTRA1 implicated in AMD susceptibility impair its capacity to regulate TGF-Î² signaling. Human Molecular Genetics, 2015, 24, 6361-6373.	1.4	42
31	Costâ€effective molecular inversion probeâ€based ABCA4 sequencing reveals deepâ€intronic variants in Stargardt disease. Human Mutation, 2019, 40, 1749-1759.	1.1	39
32	A transcriptome-wide association study based on 27 tissues identifies 106 genes potentially relevant for disease pathology in age-related macular degeneration. Scientific Reports, 2020, 10, 1584.	1.6	39
33	MonoallelicABCA4Mutations Appear Insufficient to Cause Retinopathy: A Quantitative Autofluorescence Study. , 2015, 56, 8179.		38
34	Choroidal Flow Signal in Late-Onset Stargardt Disease and Age-Related Macular Degeneration: An OCT-Angiography Study. , 2018, 59, AMD122.		38
35	Common synonymous variants in ABCA4 are protective for chloroquine induced maculopathy (toxic) Tj ETQq1 1 0.784314 rgBT /Over	0.6	36
36	The agonistic TSPO ligand XBD173 attenuates the glial response thereby protecting inner retinal neurons in a murine model of retinal ischemia. Journal of Neuroinflammation, 2019, 16, 43.	3.1	35

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37	Retinal Layer Thicknesses in Early Age-Related Macular Degeneration: Results From the German AugUR Study. , 2019, 60, 1581.		34
38	The genetics of age-related macular degeneration (AMD) â€“ Novel targets for designing treatment options?. European Journal of Pharmaceutics and Biopharmaceutics, 2015, 95, 194-202.	2.0	33
39	BEST1 protein stability and degradation pathways differ between autosomal dominant Best disease and autosomal recessive bestrophinopathy accounting for the distinct retinal phenotypes. Human Molecular Genetics, 2018, 27, 1630-1641.	1.4	32
40	Multiallelic copy number variation in the complement component 4A (C4A) gene is associated with late-stage age-related macular degeneration (AMD). Journal of Neuroinflammation, 2016, 13, 81.	3.1	31
41	On the impact of different approaches to classify age-related macular degeneration: Results from the German AugUR study. Scientific Reports, 2018, 8, 8675.	1.6	31
42	Assessment of Novel Genome-Wide Significant Gene Loci and Lesion Growth in Geographic Atrophy Secondary to Age-Related Macular Degeneration. JAMA Ophthalmology, 2019, 137, 867.	1.4	28
43	Features of Age-Related Macular Degeneration in the General Adults and Their Dependency on Age, Sex, and Smoking: Results from the German KORA Study. PLoS ONE, 2016, 11, e0167181.	1.1	27
44	Retinoschisin is linked to retinal Na/K-ATPase signaling and localization. Molecular Biology of the Cell, 2017, 28, 2178-2189.	0.9	26
45	Mutations in the Genes for Interphotoreceptor Matrix Proteoglycans, IMPG1 and IMPG2, in Patients with Vitelliform Macular Lesions. Genes, 2017, 8, 170.	1.0	24
46	Distinct Genetic Risk Profile of the Rapidly Progressing Diffuse-Trickling Subtype of Geographic Atrophy in Age-Related Macular Degeneration (AMD). , 2016, 57, 2463.		22
47	A mega-analysis of expression quantitative trait loci in retinal tissue. PLoS Genetics, 2020, 16, e1008934.	1.5	22
48	Performance of Breast Cancer Polygenic Risk Scores in 760 FemaleCHEK2Germline Mutation Carriers. Journal of the National Cancer Institute, 2021, 113, 893-899.	3.0	21
49	Mutation-Dependent Pathomechanisms Determine the Phenotype in the Bestrophinopathies. International Journal of Molecular Sciences, 2020, 21, 1597.	1.8	20
50	Evaluation of serum sphingolipids and the influence of genetic risk factors in age-related macular degeneration. PLoS ONE, 2018, 13, e0200739.	1.1	19
51	Effects of genetic variants in the TSPO gene on protein structure and stability. PLoS ONE, 2018, 13, e0195627.	1.1	19
52	Investigating the modulation of genetic effects on late AMD by age and sex: Lessons learned and two additional loci. PLoS ONE, 2018, 13, e0194321.	1.1	19
53	The Xâ€linked juvenile retinoschisis protein retinoschisin is a novel regulator of mitogenâ€activated protein kinase signalling and apoptosis in the retina. Journal of Cellular and Molecular Medicine, 2017, 21, 768-780.	1.6	18
54	Insights into the loss of the Y chromosome with age in control individuals and in patients with age-related macular degeneration using genotyping microarray data. Human Genetics, 2020, 139, 401-407.	1.8	18

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55	Learning from Fifteen Years of Genome-Wide Association Studies in Age-Related Macular Degeneration. Cells, 2020, 9, 2267.	1.8	18
56	Differentiation of murine models of â€œnegative ERGâ€•by single and repetitive light stimuli. Documenta Ophthalmologica, 2016, 132, 101-109.	1.0	17
57	Long-Term Macular Changes in the First Proband of Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC) Due to a Newly Identified Mutation inBEST1. Ophthalmic Genetics, 2016, 37, 102-108.	0.5	16
58	Pathomechanism of mutated and secreted retinoschisin in X-linked juvenile retinoschisis. Experimental Eye Research, 2018, 177, 23-34.	1.2	14
59	Altered Protein Function Caused by AMD-associated Variant rs704 Links Vitronectin to Disease Pathology. , 2020, 61, 2.		14
60	Identification of the retinoschisin-binding site on the retinal Na/K-ATPase. PLoS ONE, 2019, 14, e0216320.	1.1	13
61	Investigating the effects of additional truncating variants in DNA-repair genes on breast cancer risk in BRCA1-positive women. BMC Cancer, 2019, 19, 787.	1.1	10
62	Views of ophthalmologists on the genetics of age-related macular degeneration: Results of a qualitative study. PLoS ONE, 2018, 13, e0209328.	1.1	9
63	Clinical Heterogeneity in Autosomal Recessive Bestrophinopathy with Biallelic Mutations in the BEST1 Gene. International Journal of Molecular Sciences, 2020, 21, 9353.	1.8	8
64	A Circulating MicroRNA Profile in a Laser-Induced Mouse Model of Choroidal Neovascularization. International Journal of Molecular Sciences, 2020, 21, 2689.	1.8	8
65	Incidence, progression and risk factors of age-related macular degeneration in 35â€“95-year-old individuals from three jointly designed German cohort studies. BMJ Open Ophthalmology, 2022, 7, e000912.	0.8	7
66	Twenty-year follow-up of a familial case of PTH1R-associated primary failure of tooth eruption. American Journal of Orthodontics and Dentofacial Orthopedics, 2017, 151, 598-606.	0.8	6
67	In-Depth Functional Diagnostics of Mouse Models by Single-Flash and Flicker Electroretinograms without Adapting Background Illumination. Advances in Experimental Medicine and Biology, 2016, 854, 619-625.	0.8	5
68	The Y227N mutation affects bestrophin-1 protein stability and impairs sperm function in a mouse model of Best vitelliform macular dystrophy. Biology Open, 2019, 8, .	0.6	5
69	On the differences between megaâ€•and metaâ€•imputation and analysis exemplified on the genetics of ageâ€•related macular degeneration. Genetic Epidemiology, 2019, 43, 559-576.	0.6	5
70	Pleiotropic Locus 15q24.1 Reveals a Gender-Specific Association with Neovascular but Not Atrophic Age-Related Macular Degeneration (AMD). Cells, 2020, 9, 2257.	1.8	5
71	Epistatic interactions of genetic loci associated with age-related macular degeneration. Scientific Reports, 2021, 11, 13114.	1.6	4
72	Assigning Co-Regulated Human Genes and Regulatory Gene Clusters. Cells, 2021, 10, 2395.	1.8	4

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73	Vitronectin and Its Interaction with PAI-1 Suggests a Functional Link to Vascular Changes in AMD Pathobiology. Cells, 2022, 11, 1766.	1.8	2
74	Pleiotropic Effects of Risk Factors in Age-Related Macular Degeneration and Seemingly Unrelated Complex Diseases. Advances in Experimental Medicine and Biology, 2018, 1074, 247-255.	0.8	1
75	Correlating Adaptive Optics Images to Clinical Findings in Juvenile Macular Dystrophy with Hypotrichosis in Siblings with Homozygous <i>CDH3</i> Pathogenic Variation. Ophthalmic Research, 2020, 63, 141-151.	1.0	1
76	OUP accepted manuscript. Human Molecular Genetics, 2021, , .	1.4	1