Rando Allikmets

List of Publications by Year
in descending order

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105 papers	8,643 citations	101384 36 h-index	64668 79 g-index
108 all docs	108 docs citations	108 times ranked	7924 citing authors

#	Article	IF	Citations
1	A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Starqardt macular dystrophy. Nature Genetics, 1997, 15, 236-246.	9.4	1,277
2	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
3	Retinitis pigmentosa caused by a homozygous mutation in the Stargardt disease gene ABCR. Nature Genetics, 1998, 18, 11-12.	9.4	382
4	A 5-bp deletion in ELOVL4 is associated with two related forms of autosomal dominant macular dystrophy. Nature Genetics, 2001, 27, 89-93.	9.4	370
5	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
6	Further Evidence for an Association of ABCR Alleles with Age-Related Macular Degeneration. American Journal of Human Genetics, 2000, 67, 487-491.	2.6	287
7	Genotype/Phenotype Analysis of a Photoreceptor-Specific ATP-Binding Cassette Transporter Gene, ABCR, in Stargardt Disease. American Journal of Human Genetics, 1999, 64, 422-434.	2.6	277
8	Extended haplotypes in the complement factor H (CFH) and CFHâ€related (CFHR) family of genes protect against ageâ€related macular degeneration: Characterization, ethnic distribution and evolutionary implications. Annals of Medicine, 2006, 38, 592-604.	1.5	217
9	Clinical spectrum, genetic complexity and therapeutic approaches for retinal disease caused by ABCA4 mutations. Progress in Retinal and Eye Research, 2020, 79, 100861.	7.3	173
10	Serine and Lipid Metabolism in Macular Disease and Peripheral Neuropathy. New England Journal of Medicine, 2019, 381, 1422-1433.	13.9	166
11	Quantitative Fundus Autofluorescence in Recessive Stargardt Disease. , 2014, 55, 2841.		160
12	Genotyping Microarray (Disease Chip) for Leber Congenital Amaurosis: Detection of Modifier Alleles. , 2005, 46, 3052.		153
13	Isolation and characterization of a retinal pigment epithelial cell fluorophore: An all-trans-retinal dimer conjugate. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 7091-7096.	3.3	147
14	Clinical and Molecular Characteristics ofÂChildhood-Onset Stargardt Disease. Ophthalmology, 2015, 122, 326-334.	2.5	146
15	Frequent hypomorphic alleles account for a significant fraction of ABCA4 disease and distinguish it from age-related macular degeneration. Journal of Medical Genetics, 2017, 54, 404-412.	1.5	140
16	Analysis of theABCA4Gene by Next-Generation Sequencing. , 2011, 52, 8479.		133
17	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	1.1	118
18	Analysis of the ABCA4 genomic locus in Stargardt disease. Human Molecular Genetics, 2014, 23, 6797-6806.	1.4	117

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19	Cadherin 5 is Regulated by Corticosteroids and Associated with Central Serous Chorioretinopathy. Human Mutation, 2014, 35, 859-867.	1.1	107
20	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. Nature Genetics, 2017, 49, 559-567.	9.4	105
21	Identification and Rescue of Splice Defects Caused by Two Neighboring Deep-Intronic ABCA4 Mutations Underlying Stargardt Disease. American Journal of Human Genetics, 2018, 102, 517-527.	2.6	105
22	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	2.6	105
23	Flecks in Recessive Stargardt Disease: Short-Wavelength Autofluorescence, Near-Infrared Autofluorescence, and Optical Coherence Tomography. , 2015, 56, 5029.		93
24	G1961E mutant allele in the Stargardt disease gene ABCA4 causes bull's eye maculopathy. Experimental Eye Research, 2009, 89, 16-24.	1.2	90
25	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2012, 53, 4458.		81
26	Quantitative Fundus Autofluorescence Distinguishes ABCA4-Associated and Nonâ€“ABCA4-Associated Bull's-Eye Maculopathy. Ophthalmology, 2015, 122, 345-355.	2.5	75
27	Mapping the cis -regulatory architecture of the human retina reveals noncoding genetic variation in disease. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 9001-9012.	3.3	72
28	Outcome of ABCA4 Disease-Associated Alleles in Autosomal Recessive Retinal Dystrophies. Ophthalmology, 2013, 120, 2332-2337.	2.5	71
29	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	1.6	71
30	Correlations Among Near-Infrared and Short-Wavelength Autofluorescence and Spectral-Domain Optical Coherence Tomography in Recessive Stargardt Disease. Investigative Ophthalmology and Visual Science, 2014, 55, 8134-8143.	3.3	69
31	Whole Exome Sequencing Identifies CRB1 Defect in an Unusual Maculopathy Phenotype. Ophthalmology, 2014, 121, 1773-1782.	2.5	62
32	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
33	Extremely hypomorphic and severe deep intronic variants in the ABCA4 locus result in varying Stargardt disease phenotypes. Journal of Physical Education and Sports Management, 2018, 4, a002733.	0.5	61
34	Quantitative Fundus Autofluorescence and Optical Coherence Tomography inPRPH2/RDS-andABCA4-Associated Disease Exhibiting Phenotypic Overlap. , 2015, 56, 3159.		56
35	Quantifying Fundus Autofluorescence in Patients With Retinitis Pigmentosa. , 2017, 58, 1843.		56
36	The External Limiting Membrane in Early-Onset Stargardt Disease. , 2014, 55, 6139.		54

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37	Mutational scanning of the ABCR gene with double-gradient denaturing-gradient gel electrophoresis (DG-DGGE) in Italian Stargardt disease patients. Human Genetics, 2001, 109, 326-338.	1.8	52
38	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	1.4	52
39	Glaucomatous Optic Neuropathy Associated with Nocturnal Dip in Blood Pressure. Ophthalmology, 2018, 125, 807-814.	2.5	52
40	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. European Journal of Human Genetics, 2002, 10, 197-203.	1.4	45
41	Quantification of Peripapillary Sparing and Macular Involvement in Stargardt Disease (STGD1). , 2011, 52, 8006.		45
42	The human ATPâ€binding cassette (ABC) transporter superfamily. Human Mutation, 2022, 43, 1162-1182.	1.1	45
43	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
44	Genetic and Clinical Analysis of <scp>ABCA</scp> 4 â€Associated Disease in African American Patients. Human Mutation, 2014, 35, 1187-1194.	1.1	42
45	Near-Infrared Autofluorescence: Its Relationship to Short-Wavelength Autofluorescence and Optical Coherence Tomography in Recessive Stargardt Disease. , 2015, 56, 3226.		40
46	Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19.	1.8	39
47	The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. Ophthalmology, 2018, 125, 89-99.	2.5	39
48	CRB1Heterozygotes with Regional Retinal Dysfunction: Implications for Genetic Testing of Leber Congenital Amaurosis. , 2006, 47, 3736.		36
49	Photoreceptor cells as a source of fundus autofluorescence in recessive Stargardt disease. Journal of Neuroscience Research, 2019, 97, 98-106.	1.3	36
50	Choroidal and Retinal Thickening in Severe Preeclampsia. , 2014, 55, 5723.		35
51	Identification of a Potential Susceptibility Locus for Macular Telangiectasia Type 2. PLoS ONE, 2012, 7, e24268.	1.1	35
52	Generalized Choriocapillaris Dystrophy, a Distinct Phenotype in the Spectrum ofABCA4-Associated Retinopathies. , 2014, 55, 2766.		33
53	Serine biosynthesis defect due to haploinsufficiency of PHGDH causes retinal disease. Nature Metabolism, 2021, 3, 366-377.	5.1	32
54	Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. European Journal of Human Genetics, 2017, 25, 735-743.	1.4	31

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55	Structural and Genetic Assessment of theABCA4-Associated Optical Gap Phenotype. , 2014, 55, 7217.		30
56	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.	1.4	30
57	Gene Therapy of ABCA4-Associated Diseases. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a017301-a017301.	2.9	30
58	A non-retinoid antagonist of retinol-binding protein 4 rescues phenotype in a model of Stargardt disease without inhibiting the visual cycle. Journal of Biological Chemistry, 2018, 293, 11574-11588.	1.6	30
59	Distinct Characteristics of Inferonasal Fundus Autofluorescence Patterns in Stargardt Disease and Retinitis Pigmentosa. , 2013, 54, 6820.		29
60	Quantitative Fundus Autofluorescence and Optical Coherence Tomography inABCA4Carriers. , 2015, 56, 7274.		28
61	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	1.4	28
62	Abnormality in the external limiting membrane in early Stargardt Disease. Ophthalmic Genetics, 2013, 34, 75-77.	0.5	27
63	A caseâ€“control collapsing analysis identifies retinal dystrophy genes associated with ophthalmic disease in patients with no pathogenic ABCA4 variants. Genetics in Medicine, 2019, 21, 2336-2344.	1.1	27
64	Identification of genetic factors influencing metabolic dysregulation and retinal support for MacTel, a retinal disorder. Communications Biology, 2021, 4, 274.	2.0	26
65	A Comparison of En Face Optical Coherence Tomography and Fundus Autofluorescence in Stargardt Disease. , 2017, 58, 5227.		25
66	Mutations inGPR143/OA1andABCA4Inform Interpretations of Short-Wavelength and Near-Infrared Fundus Autofluorescence. , 2018, 59, 2459.		25
67	Cis-acting modifiers in the ABCA4 locus contribute to the penetrance of the major disease-causing variant in Stargardt disease. Human Molecular Genetics, 2021, 30, 1293-1304.	1.4	25
68	PSYCHOPHYSICAL MEASUREMENT OF ROD AND CONE THRESHOLDS IN STARGARDT DISEASE WITH FULL-FIELD STIMULI. Retina, 2014, 34, 1888-1895.	1.0	23
69	Peripapillary sparing in RDH12-associated Leber congenital amaurosis. Ophthalmic Genetics, 2017, 38, 575-579.	0.5	23
70	A Distinct Phenotype of Eyes Shut Homolog (EYS)-Retinitis Pigmentosa Is Associated With Variants Near the C-Terminus. American Journal of Ophthalmology, 2018, 190, 99-112.	1.7	23
71	Spectrum of Disease Severity and Phenotype in Choroideremia Carriers. American Journal of Ophthalmology, 2019, 207, 77-86.	1.7	21
72	Identifying and characterizing a five-gene cluster of ATP-binding cassette transporters mapping to human chromosome 17q24: a new subgroup within the ABCA subfamily. GeneScreen, 2001, 1, 157-164.	0.7	20

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73	Objective Analysis of Hyperreflective Outer Retinal Bands Imaged by Optical Coherence Tomography in Patients With Stargardt Disease. , 2015, 56, 4662.		20
74	Fine central macular dots associated with childhoodâ€onset Stargardt Disease. Acta Ophthalmologica, 2014, 92, e157-9.	0.6	17
75	Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375.	0.5	17
76	Multi-platform imaging in ABCA4-Associated Disease. Scientific Reports, 2019, 9, 6436.	1.6	17
77	Functional Analysis of Retinal Flecks in Stargardt Disease. Journal of Clinical & Experimental Ophthalmology, 2012, 03, .	0.1	17
78	A genotype-phenotype correlation matrix for ABCA4 disease based on long-term prognostic outcomes. JCI Insight, 2022, 7, .	2.3	16
79	Recessive Stargardt disease phenocopying hydroxychloroquine retinopathy. Graefe's Archive for Clinical and Experimental Ophthalmology, 2016, 254, 865-872.	1.0	15
80	CLINICAL CHARACTERIZATION OF STARGARDT DISEASE PATIENTS WITH THE p.N1868I ABCA4 MUTATION. Retina, 2019, 39, 2311-2325.	1.0	15
81	Familial discordance in Stargardt disease. Molecular Vision, 2012, 18, 227-33.	1.1	15
82	Multimodal analysis of the Preferred Retinal Location and the Transition Zone in patients with Stargardt Disease. Graefe's Archive for Clinical and Experimental Ophthalmology, 2017, 255, 1307-1317.	1.0	14
83	Clinical and Genetic Misdiagnosis of Autosomal Recessive Bestrophinopathy. JAMA Ophthalmology, 2013, 131, 1651.	1.4	11
84	Characteristic Ocular Features in Cases of Autosomal Recessive PROM1 Cone-Rod Dystrophy. , 2019, 60, 2347.		11
85	Penetrance of theABCA4p.Asn1868Ile Allele in Stargardt Disease. , 2018, 59, 5564.		10
86	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. American Journal of Ophthalmology, 2018, 195, 16-25.	1.7	10
87	Retinal Pigment Epithelium Atrophy in Recessive Stargardt Disease as Measured by Short-Wavelength and Near-Infrared Autofluorescence. Translational Vision Science and Technology, 2021, 10, 3.	1.1	9
88	Optic neuropathy and congenital glaucoma associated with probable Zika virus infection in Venezuelan patients. JMM Case Reports, 2018, 5, e005145.	1.3	9
89	Non-congenital severe ocular complications of Zika virus infection. JMM Case Reports, 2018, 5, e005152.	1.3	9
90	Late-onset pattern macular dystrophy mimicking ABCA4 and PRPH2 disease is caused by a homozygous frameshift mutation in ROM1. Journal of Physical Education and Sports Management, 2019, 5, a003624.	0.5	8

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91	Hyperautofluorescent Dots are Characteristic in Ceramide Kinase Like-associated Retinal Degeneration. Scientific Reports, 2019, 9, 876.	1.6	8
92	Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease. PLoS Genetics, 2022, 18, e1010129.	1.5	8
93	Simultaneous Expression of ABCA4 and GPR143 Mutations: A Complex Phenotypic Manifestation. , 2016, 57, 3409.		6
94	Progressive Choriocapillaris Impairment in ABCA4 Maculopathy Is Secondary to Retinal Pigment Epithelium Atrophy. , 2020, 61, 13.		5
95	Shared Features in Retinal Disorders With Involvement of Retinal Pigment Epithelium. , 2021, 62, 15.		5
96	A mutation in CRX causing pigmented paravenous retinochoroidal atrophy. European Journal of Ophthalmology, 2022, 32, NP235-NP239.	0.7	4
97	Optical Gap Biomarker in Cone-Dominant Retinal Dystrophy. American Journal of Ophthalmology, 2020, 218, 40-53.	1.7	4
98	Genetics, phenotypes, mechanisms and treatments for Leber congenital amaurosis: a paradigm shift. Expert Review of Ophthalmology, 2008, 3, 397-415.	0.3	3
99	Reevaluating the Association of Sex With ABCA4 Alleles in Patients With Stargardt Disease. JAMA Ophthalmology, 2021, 139, 654.	1.4	3
100	Expanding the phenotype of TTLL5-associated retinal dystrophy: a case series. Orphanet Journal of Rare Diseases, 2022, 17, 146.	1.2	3
101	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1
102	Comparisons Among Optical Coherence Tomography and Fundus Autofluorescence Modalities as Measurements of Atrophy in ABCA4-Associated Disease. Translational Vision Science and Technology, 2022, 11, 36.	1.1	1
103	Longitudinal Analysis of a Resolving Foveomacular Vitelliform Lesion in ABCA4 Disease. Ophthalmology Retina, 2022, 6, 847-860.	1.2	1
104	Quantitative Autofluorescence andABCA4Disease. , 2016, 57, 3297.		0
105	Transplantation of Reprogrammed Embryonic Stem Cells Improves Visual Function in a Mouse Model for Retinitis Pigmentosa. Annals of Neurosciences, 2010, 17, 185-6.	0.9	0