Jana Zernant

List of Publications by Year
in descending order

38 papers	1,965 citations	516710 16 h-index	552781 26 g-index
38 all docs	38 docs citations	38 times ranked	1964 citing authors

#	Article	IF	Citations
1	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.	3.8	217
2	Genotyping Microarray (Disease Chip) for Leber Congenital Amaurosis: Detection of Modifier Alleles. , 2005, 46, 3052.		153
3	Clinical and Molecular Characteristics ofÂChildhood-Onset Stargardt Disease. Ophthalmology, 2015, 122, 326-334.	5.2	146
4	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.	3.2	140
5	Analysis of theABCA4Gene by Next-Generation Sequencing. , 2011, 52, 8479.		133
6	In SilicoFunctional Meta-Analysis of 5,962ABCA4Variants in 3,928 Retinal Dystrophy Cases. Human Mutation, 2017, 38, 400-408.	2.5	118
7	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. Nature Genetics, 2017, 49, 559-567.	21.4	105
8	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.	6.2	105
9	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2012, 53, 4458.		81
10	Quantitative Fundus Autofluorescence Distinguishes ABCA4-Associated and Nonâ€“ABCA4-Associated Bull's-Eye Maculopathy. Ophthalmology, 2015, 122, 345-355.	5.2	75
11	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
12	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.	1.2	61
13	Quantitative Fundus Autofluorescence and Optical Coherence Tomography inPRPH2/RDS-andABCA4-Associated Disease Exhibiting Phenotypic Overlap. , 2015, 56, 3159.		56
14	The External Limiting Membrane in Early-Onset Stargardt Disease. , 2014, 55, 6139.		54
15	Genetic and Clinical Analysis of <scp>ABCA</scp> 4 â€Associated Disease in African American Patients. Human Mutation, 2014, 35, 1187-1194.	2.5	42
16	Near-Infrared Autofluorescence: Its Relationship to Short-Wavelength Autofluorescence and Optical Coherence Tomography in Recessive Stargardt Disease. , 2015, 56, 3226.		40
17	Complex inheritance of ABCA4 disease: four mutations in a family with multiple macular phenotypes. Human Genetics, 2016, 135, 9-19.	3.8	39
18	The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. Ophthalmology, 2018, 125, 89-99.	5.2	39

#	Article	IF	Citations
19	Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. European Journal of Human Genetics, 2017, 25, 735-743.	2.8	31
20	Structural and Genetic Assessment of theABCA4-Associated Optical Gap Phenotype. , 2014, 55, 7217.		30
21	Quantitative Fundus Autofluorescence and Optical Coherence Tomography inABCA4Carriers. , 2015, 56, 7274.		28
22	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	2.5	28
23	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.	2.9	25
24	Objective Analysis of Hyperreflective Outer Retinal Bands Imaged by Optical Coherence Tomography in Patients With Stargardt Disease. , 2015, 56, 4662.		20
25	Modification of the PROM1 disease phenotype by a mutation in ABCA4. Ophthalmic Genetics, 2019, 40, 369-375.	1.2	17
26	Functional Analysis of Retinal Flecks in Stargardt Disease. Journal of Clinical & Experimental Ophthalmology, 2012, 03, .	0.1	17
27	Recessive Stargardt disease phenocopying hydroxychloroquine retinopathy. Graefe's Archive for Clinical and Experimental Ophthalmology, 2016, 254, 865-872.	1.9	15
28	CLINICAL CHARACTERIZATION OF STARGARDT DISEASE PATIENTS WITH THE p.N1868I ABCA4 MUTATION. Retina, 2019, 39, 2311-2325.	1.7	15
29	Characteristic Ocular Features in Cases of Autosomal Recessive PROM1 Cone-Rod Dystrophy. , 2019, 60, 2347.		11
30	Penetrance of theABCA4p.Asn1868Ile Allele in Stargardt Disease. , 2018, 59, 5564.		10
31	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. American Journal of Ophthalmology, 2018, 195, 16-25.	3.3	10
32	Quantitative Autofluorescence as a Clinical Tool for Expedited Differential Diagnosis of Retinal Degeneration. JAMA Ophthalmology, 2015, 133, 219.	2.5	8
33	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.	1.2	8
34	Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease. PLoS Genetics, 2022, 18, e1010129.	3.5	8
35	Simultaneous Expression of ABCA4 and GPR143 Mutations: A Complex Phenotypic Manifestation. , 2016, 57, 3409.		6
36	Reevaluating the Association of Sex With ABCA4 Alleles in Patients With Stargardt Disease. JAMA Ophthalmology, 2021, 139, 654.	2.5	3

#	Article	IF	Citations
37	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.	2.2	1
38	Longitudinal Analysis of a Resolving Foveomacular Vitelliform Lesion in ABCA4 Disease. Ophthalmology Retina, 2022, 6, 847-860.	2.4	1