

Jana Zernant

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,965
citations

516215

16
h-index

552369

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 CFH-related (CFHR) family of genes protect against age-related macular degeneration: Characterization, ethnic distribution and evolutionary implications. <i>Annals of Medicine</i> , 2006, 38, 592-604.	1.5	217
2	Genotyping Microarray (Disease Chip) for Leber Congenital Amaurosis: Detection of Modifier Alleles. , 2005, 46, 3052.		153
3	Clinical and Molecular Characteristics of Childhood-Onset Stargardt Disease. <i>Ophthalmology</i> , 2015, 122, 326-334.	2.5	146
4	Frequent hypomorphic alleles account for a significant fraction of ABCA4 disease and distinguish it from age-related macular degeneration. <i>Journal of Medical Genetics</i> , 2017, 54, 404-412.	1.5	140
5	Analysis of the <i>ABCA4</i> Gene by Next-Generation Sequencing. , 2011, 52, 8479.		133
6	In Silico Functional Meta-Analysis of 5,962 <i>ABCA4</i> Variants in 3,928 Retinal Dystrophy Cases. <i>Human Mutation</i> , 2017, 38, 400-408.	1.1	118
7	Genome-wide analyses identify common variants associated with macular telangiectasia type 2. <i>Nature Genetics</i> , 2017, 49, 559-567.	9.4	105
8	Identification and Rescue of Splice Defects Caused by Two Neighboring Deep-Intronic ABCA4 Mutations Underlying Stargardt Disease. <i>American Journal of Human Genetics</i> , 2018, 102, 517-527.	2.6	105
9	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in the <i>ABCA4</i> Gene. , 2012, 53, 4458.		81
10	Quantitative Fundus Autofluorescence Distinguishes ABCA4-Associated and Non-ABCA4-Associated Bull's-Eye Maculopathy. <i>Ophthalmology</i> , 2015, 122, 345-355.	2.5	75
11	Correlations Among Near-Infrared and Short-Wavelength Autofluorescence and Spectral-Domain Optical Coherence Tomography in Recessive Stargardt Disease. <i>Investigative Ophthalmology and Visual Science</i> , 2014, 55, 8134-8143.	3.3	69
12	Extremely hypomorphic and severe deep intronic variants in the <i>ABCA4</i> locus result in varying Stargardt disease phenotypes. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a002733.	0.5	61
13	Quantitative Fundus Autofluorescence and Optical Coherence Tomography in <i>PRPH2/RDS</i> - and <i>ABCA4</i> -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 <i>ABCA4</i> -Associated Disease in African American Patients. <i>Human Mutation</i> , 2014, 35, 1187-1194.	1.1	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. <i>Human Genetics</i> , 2016, 135, 9-19.	1.8	39
18	The Rapid-Onset Chorioretinopathy Phenotype of ABCA4 Disease. <i>Ophthalmology</i> , 2018, 125, 89-99.	2.5	39

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19	Genotypic spectrum and phenotype correlations of ABCA4-associated disease in patients of south Asian descent. <i>European Journal of Human Genetics</i> , 2017, 25, 735-743.	1.4	31
20	Structural and Genetic Assessment of the ABCA4-Associated Optical Gap Phenotype. , 2014, 55, 7217.		30
21	Quantitative Fundus Autofluorescence and Optical Coherence Tomography in ABCA4 Carriers. , 2015, 56, 7274.		28
22	New Mutations in the <i>RAB28</i> Gene in 2 Spanish Families With Cone-Rod Dystrophy. <i>JAMA Ophthalmology</i> , 2015, 133, 133.	1.4	28
23	<i>Cis</i> -acting modifiers in the <i>ABCA4</i> locus contribute to the penetrance of the major disease-causing variant in Stargardt disease. <i>Human Molecular Genetics</i> , 2021, 30, 1293-1304.	1.4	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 <i>PROM1</i> disease phenotype by a mutation in <i>ABCA4</i> . <i>Ophthalmic Genetics</i> , 2019, 40, 369-375.	0.5	17
26	Functional Analysis of Retinal Flecks in Stargardt Disease. <i>Journal of Clinical & Experimental Ophthalmology</i> , 2012, 03, .	0.1	17
27	Recessive Stargardt disease phenocopying hydroxychloroquine retinopathy. <i>Graefes Archive for Clinical and Experimental Ophthalmology</i> , 2016, 254, 865-872.	1.0	15
28	CLINICAL CHARACTERIZATION OF STARGARDT DISEASE PATIENTS WITH THE p.N1868I ABCA4 MUTATION. <i>Retina</i> , 2019, 39, 2311-2325.	1.0	15
29	Characteristic Ocular Features in Cases of Autosomal Recessive <i>PROM1</i> Cone-Rod Dystrophy. , 2019, 60, 2347.		11
30	Penetrance of the <i>ABCA4</i> p.Asn1868Ile Allele in Stargardt Disease. , 2018, 59, 5564.		10
31	Deep Scleral Exposure: A Degenerative Outcome of End-Stage Stargardt Disease. <i>American Journal of Ophthalmology</i> , 2018, 195, 16-25.	1.7	10
32	Quantitative Autofluorescence as a Clinical Tool for Expedited Differential Diagnosis of Retinal Degeneration. <i>JAMA Ophthalmology</i> , 2015, 133, 219.	1.4	8
33	Late-onset pattern macular dystrophy mimicking <i>ABCA4</i> and <i>PRPH2</i> disease is caused by a homozygous frameshift mutation in <i>ROM1</i> . <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003624.	0.5	8
34	Rare and common variants in <i>ROM1</i> and <i>PRPH2</i> genes trans-modify Stargardt/ABCA4 disease. <i>PLoS Genetics</i> , 2022, 18, e1010129.	1.5	8
35	Simultaneous Expression of ABCA4 and GPR143 Mutations: A Complex Phenotypic Manifestation. , 2016, 57, 3409.		6
36	Reevaluating the Association of Sex With <i>ABCA4</i> Alleles in Patients With Stargardt Disease. <i>JAMA Ophthalmology</i> , 2021, 139, 654.	1.4	3

#	ARTICLE	IF	CITATIONS
37	Comparisons Among Optical Coherence Tomography and Fundus Autofluorescence Modalities as Measurements of Atrophy in <i>ABCA4</i> -Associated Disease. <i>Translational Vision Science and Technology</i> , 2022, 11, 36.	1.1	1
38	Longitudinal Analysis of a Resolving Foveomacular Vitelliform Lesion in <i>ABCA4</i> Disease. <i>Ophthalmology Retina</i> , 2022, 6, 847-860.	1.2	1