

Rob W J Collin

List of Publications by Year in descending order

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Version: 2024-02-01

102
papers

6,431
citations

76326

40
h-index

76900

74
g-index

104
all docs

104
docs citations

104
times ranked

6358
citing authors

#	ARTICLE	IF	CITATIONS
1	Non-syndromic retinitis pigmentosa. <i>Progress in Retinal and Eye Research</i> , 2018, 66, 157-186.	15.5	565
2	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. <i>Nature Genetics</i> , 2010, 42, 840-850.	21.4	295
3	Next-generation genetic testing for retinitis pigmentosa. <i>Human Mutation</i> , 2012, 33, 963-972.	2.5	258
4	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. <i>American Journal of Human Genetics</i> , 2010, 86, 240-247.	6.2	202
5	Homozygosity Mapping Reveals PDE6C Mutations in Patients with Early-Onset Cone Photoreceptor Disorders. <i>American Journal of Human Genetics</i> , 2009, 85, 240-247.	6.2	194
6	Delivery of oligonucleotide-based therapeutics: challenges and opportunities. <i>EMBO Molecular Medicine</i> , 2021, 13, e13243.	6.9	181
7	Clinical spectrum, genetic complexity and therapeutic approaches for retinal disease caused by ABCA4 mutations. <i>Progress in Retinal and Eye Research</i> , 2020, 79, 100861.	15.5	173
8	Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. <i>Genetics in Medicine</i> , 2019, 21, 1751-1760.	2.4	147
9	<i>ZNF408</i> is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 9856-9861.	7.1	144
10	Identification of a 2 Mb Human Ortholog of <i>Drosophila</i> eyes shut/spacemaker that Is Mutated in Patients with Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2008, 83, 594-603.	6.2	141
11	Mutations in IFT172 cause isolated retinal degeneration and Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 230-242.	2.9	136
12	<i>ABCA4</i> midgenes reveal the full splice spectrum of all reported noncanonical splice site variants in Stargardt disease. <i>Genome Research</i> , 2018, 28, 100-110.	5.5	134
13	Splice-Modulating Oligonucleotide QR-110 Restores CEP290 mRNA and Function in Human c.2991+1655A>G LCA10 Models. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 12, 730-740.	5.1	130
14	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. <i>American Journal of Human Genetics</i> , 2008, 82, 125-138.	6.2	127
15	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. <i>Human Mutation</i> , 2010, 31, 656-666.	2.5	126
16	Antisense Oligonucleotide (AON)-based Therapy for Leber Congenital Amaurosis Caused by a Frequent Mutation in CEP290. <i>Molecular Therapy - Nucleic Acids</i> , 2012, 1, e14.	5.1	116
17	Mutations in SPATA7 Cause Leber Congenital Amaurosis and Juvenile Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2009, 84, 380-387.	6.2	111
18	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. <i>Genetics in Medicine</i> , 2019, 21, 1761-1771.	2.4	111

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19	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.	6.2	105
20	Antisense Oligonucleotide-based Splice Correction for USH2A-associated Retinal Degeneration Caused by a Frequent Deep-intronic Mutation. <i>Molecular Therapy - Nucleic Acids</i> , 2016, 5, e381.	5.1	104
21	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2010, 87, 199-208.	6.2	98
22	Photoreceptor Progenitor mRNA Analysis Reveals Exon Skipping Resulting from the ABCA4 c.5461-10Tâ†’C Mutation in Stargardt Disease. <i>Ophthalmology</i> , 2016, 123, 1375-1385.	5.2	96
23	Exome Sequencing and cis-Regulatory Mapping Identify Mutations in MAK, a Gene Encoding a Regulator of Ciliary Length, as a Cause of Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2011, 89, 253-264.	6.2	95
24	Homozygosity Mapping in Patients with Coneâ€™Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
25	In vitro and in vivo rescue of aberrant splicing in CEP290-associated LCA by antisense oligonucleotide delivery. <i>Human Molecular Genetics</i> , 2016, 25, ddw118.	2.9	92
26	Mutations in C2ORF71 Cause Autosomal-Recessive Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2010, 86, 783-788.	6.2	88
27	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. <i>American Journal of Human Genetics</i> , 2012, 90, 102-109.	6.2	82
28	The pros and cons of vertebrate animal models for functional and therapeutic research on inherited retinal dystrophies. <i>Progress in Retinal and Eye Research</i> , 2015, 48, 137-159.	15.5	81
29	Defects in the Cell Signaling Mediator Î²-Catenin Cause the Retinal Vascular Condition FEVR. <i>American Journal of Human Genetics</i> , 2017, 100, 960-968.	6.2	74
30	Mutations in the EYS Gene Account for Approximately 5% of Autosomal Recessive Retinitis Pigmentosa and Cause a Fairly Homogeneous Phenotype. <i>Ophthalmology</i> , 2010, 117, 2026-2033.e7.	5.2	72
31	Comprehensive genotyping reveals RPE65 as the most frequently mutated gene in Leber congenital amaurosis in Denmark. <i>European Journal of Human Genetics</i> , 2016, 24, 1071-1079.	2.8	69
32	High-Resolution Homozygosity Mapping Is a Powerful Tool to Detect Novel Mutations Causative of Autosomal Recessive RP in the Dutch Population. , 2011, 52, 2227.		67
33	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. <i>Nature Genetics</i> , 2008, 40, 1335-1340.	21.4	65
34	A Novel Nonsense Mutation in <i>CEP290</i> Induces Exon Skipping and Leads to a Relatively Mild Retinal Phenotype. , 2010, 51, 3646.		65
35	Missense mutations in <i>POU4F3</i> cause autosomal dominant hearing impairment DFNA15 and affect subcellular localization and DNA binding. <i>Human Mutation</i> , 2008, 29, 545-554.	2.5	62
36	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. <i>Ophthalmology</i> , 2011, 118, 1444-1448.	5.2	61

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37	Molecular Therapies for Inherited Retinal Diseases—Current Standing, Opportunities and Challenges. <i>Genes</i> , 2019, 10, 654.	2.4	59
38	Involvement of DFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. <i>Human Mutation</i> , 2007, 28, 718-723.	2.5	58
39	PCARE and WASF3 regulate ciliary F-actin assembly that is required for the initiation of photoreceptor outer segment disk formation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 9922-9931.	7.1	58
40	Novel Null Mutations in the <i>EYS</i> Gene Are a Frequent Cause of Autosomal Recessive Retinitis Pigmentosa in the Israeli Population. , 2010, 51, 4387.		57
41	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. <i>Molecular Therapy - Nucleic Acids</i> , 2020, 21, 412-427.	5.1	55
42	Unexpected CEP290 mRNA Splicing in a Humanized Knock-In Mouse Model for Leber Congenital Amaurosis. <i>PLoS ONE</i> , 2013, 8, e79369.	2.5	55
43	Antisense Oligonucleotide Screening to Optimize the Rescue of the Splicing Defect Caused by the Recurrent Deep-Intronic ABCA4 Variant c.4539+2001G>A in Stargardt Disease. <i>Genes</i> , 2019, 10, 452.	2.4	50
44	A missense mutation in the splicing factor gene <i>DHX38</i> is associated with early-onset retinitis pigmentosa with macular coloboma. <i>Journal of Medical Genetics</i> , 2014, 51, 444-448.	3.2	48
45	Intravitreal antisense oligonucleotide sepfarsen in Leber congenital amaurosis type 10: a phase 1b/2 trial. <i>Nature Medicine</i> , 2022, 28, 1014-1021.	30.7	46
46	Whole-exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations. <i>Human Molecular Genetics</i> , 2015, 24, 4037-4048.	2.9	41
47	<i>IMPG2</i> -Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
48	Eyes shut homolog is important for the maintenance of photoreceptor morphology and visual function in zebrafish. <i>PLoS ONE</i> , 2018, 13, e0200789.	2.5	37
49	Progressive Sensorineural Hearing Loss and Normal Vestibular Function in a Dutch DFNB7/11 Family with a Novel Mutation in <i>TMC1</i> . <i>Audiology and Neuro-Otology</i> , 2011, 16, 93-105.	1.3	36
50	A look into retinal organoids: methods, analytical techniques, and applications. <i>Cellular and Molecular Life Sciences</i> , 2021, 78, 6505-6532.	5.4	36
51	Autosomal Recessive NRL Mutations in Patients with Enhanced S-Cone Syndrome. <i>Genes</i> , 2018, 9, 68.	2.4	35
52	Mid-frequency DFNA8/12 hearing loss caused by a synonymous TECTA mutation that affects an exonic splice enhancer. <i>European Journal of Human Genetics</i> , 2008, 16, 1430-1436.	2.8	33
53	Therapeutic effects of the mitochondrial ROS-redox modulator KH176 in a mammalian model of Leigh Disease. <i>Scientific Reports</i> , 2017, 7, 11733.	3.3	33
54	Antisense Oligonucleotide-Based Splice Correction of a Deep-Intronic Mutation in CHM Underlying Choroideremia. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1074, 83-89.	1.6	33

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55	Applications of antisense oligonucleotides for the treatment of inherited retinal diseases. <i>Current Opinion in Ophthalmology</i> , 2017, 28, 260-266.	2.9	33
56	Antisense Oligonucleotide-Based Rescue of Aberrant Splicing Defects Caused by 15 Pathogenic Variants in ABCA4. <i>International Journal of Molecular Sciences</i> , 2021, 22, 4621.	4.1	30
57	Phenotype Description of a Novel DFNA9/COCH Mutation, I109T. <i>Annals of Otology, Rhinology and Laryngology</i> , 2007, 116, 349-357.	1.1	29
58	Novel compound heterozygous NMNAT1 variants associated with Leber congenital amaurosis. <i>Molecular Vision</i> , 2014, 20, 753-9.	1.1	29
59	Nonpenetrance of the Most Frequent Autosomal Recessive Leber Congenital Amaurosis Mutation in <i>NMNAT1</i> . <i>JAMA Ophthalmology</i> , 2014, 132, 1002.	2.5	28
60	Novel insights into the molecular pathogenesis of CYP4V2-associated Bietti's retinal dystrophy. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 14-29.	1.2	28
61	Molecular genetic analysis of retinitis pigmentosa in Indonesia using genome-wide homozygosity mapping. <i>Molecular Vision</i> , 2011, 17, 3013-24.	1.1	28
62	Homozygosity Mapping and Targeted Sanger Sequencing Reveal Genetic Defects Underlying Inherited Retinal Disease in Families from Pakistan. <i>PLoS ONE</i> , 2015, 10, e0119806.	2.5	27
63	Species-Dependent Splice Recognition of a Cryptic Exon Resulting from a Recurrent Intronic CEP290 Mutation that Causes Congenital Blindness. <i>International Journal of Molecular Sciences</i> , 2015, 16, 5285-5298.	4.1	27
64	A novel crumbs homolog 1 mutation in a family with retinitis pigmentosa, nanophthalmos, and optic disc drusen. <i>Molecular Vision</i> , 2012, 18, 2447-53.	1.1	27
65	Prenylation defects in inherited retinal diseases. <i>Journal of Medical Genetics</i> , 2014, 51, 143-151.	3.2	26
66	Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. <i>Molecular Vision</i> , 2013, 19, 644-53.	1.1	26
67	EYS mutation update: In silico assessment of 271 reported and 26 novel variants in patients with retinitis pigmentosa. <i>Human Mutation</i> , 2018, 39, 177-186.	2.5	23
68	Antisense Oligonucleotide-Based Splicing Correction in Individuals with Leber Congenital Amaurosis due to Compound Heterozygosity for the c.2991+1655A>G Mutation in CEP290. <i>International Journal of Molecular Sciences</i> , 2018, 19, 753.	4.1	23
69	A novel mutation in GRK1 causes Oguchi disease in a consanguineous Pakistani family. <i>Molecular Vision</i> , 2009, 15, 1788-93.	1.1	22
70	Variants in Nebulin (NEB) Are Linked to the Development of Familial Primary Angle Closure Glaucoma in Basset Hounds. <i>PLoS ONE</i> , 2015, 10, e0126660.	2.5	21
71	Mutations in <i>AGBL5</i> , Encoding β -Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
72	Clinical Characterization of 66 Patients With Congenital Retinal Disease Due to the Deep-Intronic c.2991+1655A>G Mutation in <i>CEP290</i> . , 2018, 59, 4384.		21

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73	Audiometric and Vestibular Features in a Second Dutch DFNA20/26 Family with a Novel Mutation in <i>ACTG1</i> . <i>Annals of Otology, Rhinology and Laryngology</i> , 2009, 118, 382-390.	1.1	20
74	The Molecular Basis of Retinal Dystrophies in Pakistan. <i>Genes</i> , 2014, 5, 176-195.	2.4	20
75	Antisense Oligonucleotide Therapy for Inherited Retinal Dystrophies. <i>Advances in Experimental Medicine and Biology</i> , 2016, 854, 517-524.	1.6	20
76	Clinical Characteristics of a Dutch DFNA9 Family with a Novel <i>COCH</i> Mutation, G87W. <i>Audiology and Neuro-Otology</i> , 2007, 12, 77-84.	1.3	18
77	Autosomal recessive retinitis pigmentosa with <i>RP1</i> mutations is associated with myopia. <i>British Journal of Ophthalmology</i> , 2015, 99, 1360-1365.	3.9	18
78	<i>C2orf71a/pcare1</i> is important for photoreceptor outer segment morphogenesis and visual function in zebrafish. <i>Scientific Reports</i> , 2018, 8, 9675.	3.3	18
79	The efficacy of microarray screening for autosomal recessive retinitis pigmentosa in routine clinical practice. <i>Molecular Vision</i> , 2015, 21, 461-76.	1.1	18
80	Missense mutations at homologous positions in the fourth and fifth laminin A G-like domains of eyes shut homolog cause autosomal recessive retinitis pigmentosa. <i>Molecular Vision</i> , 2010, 16, 2753-9.	1.1	17
81	Identification of recurrent and novel mutations in <i>TULP1</i> in Pakistani families with early-onset retinitis pigmentosa. <i>Molecular Vision</i> , 2012, 18, 1226-37.	1.1	17
82	Extending the Spectrum of <i>EYS</i> -Associated Retinal Disease to Macular Dystrophy. , 2019, 60, 2049.		16
83	Identification of a novel nonsense mutation in <i>RP1</i> that causes autosomal recessive retinitis pigmentosa in an Indonesian family. <i>Molecular Vision</i> , 2012, 18, 2411-9.	1.1	16
84	Identification of Novel Mutations in Pakistani Families With Autosomal Recessive Retinitis Pigmentosa. <i>JAMA Ophthalmology</i> , 2011, 129, 1377.	2.4	15
85	A homozygous p.Glu150Lys mutation in the opsin gene of two Pakistani families with autosomal recessive retinitis pigmentosa. <i>Molecular Vision</i> , 2009, 15, 2526-34.	1.1	15
86	Novel mutations in <i>RDH5</i> cause fundus albipunctatus in two consanguineous Pakistani families. <i>Molecular Vision</i> , 2012, 18, 1558-71.	1.1	15
87	Design and In Vitro Use of Antisense Oligonucleotides to Correct Pre-mRNA Splicing Defects in Inherited Retinal Dystrophies. <i>Methods in Molecular Biology</i> , 2018, 1715, 61-78.	0.9	14
88	An FEVR-associated mutation in <i>ZNF408</i> alters the expression of genes involved in the development of vasculature. <i>Human Molecular Genetics</i> , 2018, 27, 3519-3527.	2.9	14
89	<i>PRPH2</i> mutation update: In silico assessment of 245 reported and 7 novel variants in patients with retinal disease. <i>Human Mutation</i> , 2021, 42, 1521-1547.	2.5	13
90	Novel <i>CNGA3</i> and <i>CNGB3</i> mutations in two Pakistani families with achromatopsia. <i>Molecular Vision</i> , 2010, 16, 774-81.	1.1	13

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91	Biosynthesis and differential processing of two pools of amyloid- β precursor protein in a physiologically inducible neuroendocrine cell. <i>Journal of Neurochemistry</i> , 2005, 94, 1015-1024.	3.9	12
92	Genomic Approaches For the Discovery of Genes Mutated in Inherited Retinal Degeneration. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2014, 4, a017137-a017137.	6.2	10
93	P-cadherin mutations are associated with high basal Wnt activity and stemness in canine mammary tumor cell lines. <i>Oncotarget</i> , 2019, 10, 2930-2946.	1.8	10
94	Identification and expression of the first nonmammalian amyloid-beta precursor-like protein APLP2 in the amphibian <i>Xenopus laevis</i> . <i>FEBS Journal</i> , 2004, 271, 1906-1912.	0.2	9
95	A Nonsense Mutation in <i>FAM161A</i> Is a Recurrent Founder Allele in Dutch and Belgian Individuals With Autosomal Recessive Retinitis Pigmentosa. , 2015, 56, 7418.		9
96	Detection and quantification of a KIF11 mosaicism in a subject presenting familial exudative vitreoretinopathy with microcephaly. <i>European Journal of Human Genetics</i> , 2018, 26, 1819-1823.	2.8	9
97	In or Out? New Insights on Exon Recognition through Splice-Site Interdependency. <i>International Journal of Molecular Sciences</i> , 2020, 21, 2300.	4.1	8
98	Modeling <i>ZNF408</i> -Associated FEVR in Zebrafish Results in Abnormal Retinal Vasculature. , 2020, 61, 39.		7
99	Zebrafish as a Model to Evaluate a CRISPR/Cas9-Based Exon Excision Approach as a Future Treatment Option for EYS-Associated Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9154.	4.1	6
100	Development and Use of Cellular Systems to Assess and Correct Splicing Defects. <i>Methods in Molecular Biology</i> , 2022, 2434, 145-165.	0.9	3
101	PCARE requires coiled coil, RP62 kinase-binding and EVH1 domain-binding motifs for ciliary expansion. <i>Human Molecular Genetics</i> , 2022, 31, 2560-2570.	2.9	3
102	Unraveling the mysteries of pre-mRNA splicing in the retina via stem cell technology. <i>Stem Cell Investigation</i> , 2016, 3, 72-72.	3.0	0