Rob W J Collin

List of Publications by Year
in descending order

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	Development and Use of Cellular Systems to Assess and Correct Splicing Defects. Methods in Molecular Biology, 2022, 2434, 145-165.	0.9	3
2	PCARE requires coiled coil, RP62 kinase-binding and EVH1 domain-binding motifs for ciliary expansion. Human Molecular Genetics, 2022, 31, 2560-2570.	2.9	3
3	Intravitreal antisense oligonucleotide sepofarsen in Leber congenital amaurosis type 10: a phase 1b/2 trial. Nature Medicine, 2022, 28, 1014-1021.	30.7	46
4	Delivery of oligonucleotideâ€based therapeutics: challenges and opportunities. EMBO Molecular Medicine, 2021, 13, e13243.	6.9	181
5	Antisense Oligonucleotide-Based Rescue of Aberrant Splicing Defects Caused by 15 Pathogenic Variants in ABCA4. International Journal of Molecular Sciences, 2021, 22, 4621.	4.1	30
6	A look into retinal organoids: methods, analytical techniques, and applications. Cellular and Molecular Life Sciences, 2021, 78, 6505-6532.	5.4	36
7	Zebrafish as a Model to Evaluate a CRISPR/Cas9-Based Exon Excision Approach as a Future Treatment Option for EYS-Associated Retinitis Pigmentosa. International Journal of Molecular Sciences, 2021, 22, 9154.	4.1	6
8	PRPH2 mutation update: In silico assessment of 245 reported and 7 novel variants in patients with retinal disease. Human Mutation, 2021, 42, 1521-1547.	2.5	13
9	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	5.1	55
10	Modeling ZNF408-Associated FEVR in Zebrafish Results in Abnormal Retinal Vasculature. , 2020, 61, 39.		7
11	Clinical spectrum, genetic complexity and therapeutic approaches for retinal disease caused by ABCA4 mutations. Progress in Retinal and Eye Research, 2020, 79, 100861.	15.5	173
12	PCARE and WASF3 regulate ciliary F-actin assembly that is required for the initiation of photoreceptor outer segment disk formation. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 9922-9931.	7.1	58
13	In or Out? New Insights on Exon Recognition through Splice-Site Interdependency. International Journal of Molecular Sciences, 2020, 21, 2300.	4.1	8
14	Molecular Therapies for Inherited Retinal Diseasesâ€”Current Standing, Opportunities and Challenges. Genes, 2019, 10, 654.	2.4	59
15	ABCA4-associated disease as a model for missing heritability in autosomal recessive disorders: novel noncoding splice, cis-regulatory, structural, and recurrent hypomorphic variants. Genetics in Medicine, 2019, 21, 1761-1771.	2.4	111
16	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. Genes, 2019, 10, 452.	2.4	50
17	Extending the Spectrum of EYS-Associated Retinal Disease to Macular Dystrophy. , 2019, 60, 2049.		16
18	Deep-intronic ABCA4 variants explain missing heritability in Stargardt disease and allow correction of splice defects by antisense oligonucleotides. Genetics in Medicine, 2019, 21, 1751-1760.	2.4	147

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19	P-cadherin mutations are associated with high basal Wnt activity and stemness in canine mammary tumor cell lines. Oncotarget, 2019, 10, 2930-2946.	1.8	10
20	Antisense Oligonucleotide-Based Splice Correction of a Deep-Intronic Mutation in CHM Underlying Choroideremia. Advances in Experimental Medicine and Biology, 2018, 1074, 83-89.	1.6	33
21	Non-syndromic retinitis pigmentosa. Progress in Retinal and Eye Research, 2018, 66, 157-186.	15.5	565
22	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
23	ABCA4 midigenes reveal the full splice spectrum of all reported noncanonical splice site variants in Stargardt disease. Genome Research, 2018, 28, 100-110.	5.5	134
24	Design and In Vitro Use of Antisense Oligonucleotides to Correct Pre-mRNA Splicing Defects in Inherited Retinal Dystrophies. Methods in Molecular Biology, 2018, 1715, 61-78.	0.9	14
25	EYSmutation update: In silico assessment of 271 reported and 26 novel variants in patients with retinitis pigmentosa. Human Mutation, 2018, 39, 177-186.	2.5	23
26	Clinical Characterization of 66 Patients With Congenital Retinal Disease Due to the Deep-Intronic c.2991+1655A>G Mutation inCEP290. , 2018, 59, 4384.		21
27	Detection and quantification of a KIF11 mosaicism in a subject presenting familial exudative vitreoretinopathy with microcephaly. European Journal of Human Genetics, 2018, 26, 1819-1823.	2.8	9
28	C2orf71a/pcare1 is important for photoreceptor outer segment morphogenesis and visual function in zebrafish. Scientific Reports, 2018, 8, 9675.	3.3	18
29	Eyes shut homolog is important for the maintenance of photoreceptor morphology and visual function in zebrafish. PLoS ONE, 2018, 13, e0200789.	2.5	37
30	Autosomal Recessive NRL Mutations in Patients with Enhanced S-Cone Syndrome. Genes, 2018, 9, 68.	2.4	35
31	Antisense Oligonucleotide-Based Splicing Correction in Individuals with Leber Congenital Amaurosis due to Compound Heterozygosity for the c.2991+1655A>G Mutation in CEP290. International Journal of Molecular Sciences, 2018, 19, 753.	4.1	23
32	An FEVR-associated mutation in ZNF408 alters the expression of genes involved in the development of vasculature. Human Molecular Genetics, 2018, 27, 3519-3527.	2.9	14
33	Splice-Modulating Oligonucleotide QR-110 Restores CEP290 mRNA and Function in Human c.2991+1655A>G LCA10 Models. Molecular Therapy - Nucleic Acids, 2018, 12, 730-740.	5.1	130
34	Defects in the Cell Signaling Mediator Î²-Catenin Cause the Retinal Vascular Condition FEVR. American Journal of Human Genetics, 2017, 100, 960-968.	6.2	74
35	Therapeutic effects of the mitochondrial ROS-redox modulator KH176 in a mammalian model of Leigh Disease. Scientific Reports, 2017, 7, 11733.	3.3	33
36	Applications of antisense oligonucleotides for the treatment of inherited retinal diseases. Current Opinion in Ophthalmology, 2017, 28, 260-266.	2.9	33

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37	Unraveling the mysteries of pre-mRNA splicing in the retina via stem cell technology. Stem Cell Investigation, 2016, 3, 72-72.	3.0	0
38	Mutations in AGBL5, Encoding Î±-Tubulin Deglutamylase, Are Associated With Autosomal Recessive Retinitis Pigmentosa. , 2016, 57, 6180.		21
39	In vitroandin vivorescue of aberrant splicing inCEP290-associated LCA by antisense oligonucleotide delivery. Human Molecular Genetics, 2016, 25, ddw118.	2.9	92
40	Antisense Oligonucleotide-based Splice Correction for USH2A-associated Retinal Degeneration Caused by a Frequent Deep-intronic Mutation. Molecular Therapy - Nucleic Acids, 2016, 5, e381.	5.1	104
41	Photoreceptor Progenitor mRNA Analysis Reveals Exon Skipping Resulting from the ABCA4 c.5461-10Tâ†'C Mutation in Stargardt Disease. Ophthalmology, 2016, 123, 1375-1385.	5.2	96
42	Comprehensive genotyping reveals RPE65 as the most frequently mutated gene in Leber congenital amaurosis in Denmark. European Journal of Human Genetics, 2016, 24, 1071-1079.	2.8	69
43	Antisense Oligonucleotide Therapy for Inherited Retinal Dystrophies. Advances in Experimental Medicine and Biology, 2016, 854, 517-524.	1.6	20
44	A Nonsense Mutation inFAM161AIs a Recurrent Founder Allele in Dutch and Belgian Individuals With Autosomal Recessive Retinitis Pigmentosa. , 2015, 56, 7418.		9
45	Homozygosity Mapping and Targeted Sanger Sequencing Reveal Genetic Defects Underlying Inherited Retinal Disease in Families from Pakistan. PLoS ONE, 2015, 10, e0119806.	2.5	27
46	Variants in Nebulin (NEB) Are Linked to the Development of Familial Primary Angle Closure Glaucoma in Basset Hounds. PLoS ONE, 2015, 10, e0126660.	2.5	21
47	Species-Dependent Splice Recognition of a Cryptic Exon Resulting from a Recurrent Intronic CEP290 Mutation that Causes Congenital Blindness. International Journal of Molecular Sciences, 2015, 16, 5285-5298.	4.1	27
48	Whole-exome sequencing reveals ZNF408 as a new gene associated with autosomal recessive retinitis pigmentosa with vitreal alterations. Human Molecular Genetics, 2015, 24, 4037-4048.	2.9	41
49	The pros and cons of vertebrate animal models for functional and therapeutic research on inherited retinal dystrophies. Progress in Retinal and Eye Research, 2015, 48, 137-159.	15.5	81
50	Autosomal recessive retinitis pigmentosa withRP1mutations is associated with myopia. British Journal of Ophthalmology, 2015, 99, 1360-1365.	3.9	18
51	Novel insights into the molecular pathogenesis of CYP 4V2 â€associated Bietti's retinal dystrophy. Molecular Genetics & Genomic Medicine, 2015, 3, 14-29.	1.2	28
52	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
53	The efficacy of microarray screening for autosomal recessive retinitis pigmentosa in routine clinical practice. Molecular Vision, 2015, 21, 461-76.	1.1	18
54	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37

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55	Prenylation defects in inherited retinal diseases. Journal of Medical Genetics, 2014, 51, 143-151.	3.2	26
56	Nonpenetrance of the Most Frequent Autosomal Recessive Leber Congenital Amaurosis Mutation inNMNAT1. JAMA Ophthalmology, 2014, 132, 1002.	2.5	28
57	A missense mutation in the splicing factor geneDHX38is associated with early-onset retinitis pigmentosa with macular coloboma. Journal of Medical Genetics, 2014, 51, 444-448.	3.2	48
58	Genomic Approaches For the Discovery of Genes Mutated in Inherited Retinal Degeneration. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a017137-a017137.	6.2	10
59	The Molecular Basis of Retinal Dystrophies in Pakistan. Genes, 2014, 5, 176-195.	2.4	20
60	Novel compound heterozygous NMNAT1 variants associated with Leber congenital amaurosis. Molecular Vision, 2014, 20, 753-9.	1.1	29
61	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	7.1	144
62	Unexpected CEP290 mRNA Splicing in a Humanized Knock-In Mouse Model for Leber Congenital Amaurosis. PLoS ONE, 2013, 8, e79369.	2.5	55
63	Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. Molecular Vision, 2013, 19, 644-53.	1.1	26
64	Antisense Oligonucleotide (AON)-based Therapy for Leber Congenital Amaurosis Caused by a Frequent Mutation in CEP290. Molecular Therapy - Nucleic Acids, 2012, 1, e14.	5.1	116
65	Nextâ€generation genetic testing for retinitis pigmentosa. Human Mutation, 2012, 33, 963-972.	2.5	258
66	Mutations in C8orf37, Encoding a Ciliary Protein, are Associated with Autosomal-Recessive Retinal Dystrophies with Early Macular Involvement. American Journal of Human Genetics, 2012, 90, 102-109.	6.2	82
67	Identification of recurrent and novel mutations in TULP1 in Pakistani families with early-onset retinitis pigmentosa. Molecular Vision, 2012, 18, 1226-37.	1.1	17
68	Novel mutations in RDH5 cause fundus albipunctatus in two consanguineous Pakistani families. Molecular Vision, 2012, 18, 1558-71.	1.1	15
69	A novel crumbs homolog 1 mutation in a family with retinitis pigmentosa, nanophthalmos, and optic disc drusen. Molecular Vision, 2012, 18, 2447-53.	1.1	27
70	Identification of a novel nonsense mutation in RP1 that causes autosomal recessive retinitis pigmentosa in an Indonesian family. Molecular Vision, 2012, 18, 2411-9.	1.1	16
71	CLRN1 Mutations Cause Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2011, 118, 1444-1448.	5.2	61
72	Exome Sequencing and cis-Regulatory Mapping Identify Mutations in MAK, a Gene Encoding a Regulator of Ciliary Length, as a Cause of Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 89, 253-264.	6.2	95

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73	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
74	Progressive Sensorineural Hearing Loss and Normal Vestibular Function in a Dutch DFNB7/11 Family with a Novel Mutation in <i>TMC1</i>. Audiology and Neuro-Otology, 2011, 16, 93-105.	1.3	36
75	Identification of Novel Mutations in Pakistani Families With Autosomal Recessive Retinitis Pigmentosa. JAMA Ophthalmology, 2011, 129, 1377.	2.4	15
76	Molecular genetic analysis of retinitis pigmentosa in Indonesia using genome-wide homozygosity mapping. Molecular Vision, 2011, 17, 3013-24.	1.1	28
77	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	6.2	202
78	Mutations in C2ORF71 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 783-788.	6.2	88
79	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	6.2	98
80	Overview of the mutation spectrum in familial exudative vitreoretinopathy and Norrie disease with identification of 21 novel variants in FZD4, LRP5, and NDP. Human Mutation, 2010, 31, 656-666.	2.5	126
81	Candidate exome capture identifies mutation of SDCCAG8 as the cause of a retinal-renal ciliopathy. Nature Genetics, 2010, 42, 840-850.	21.4	295
82	Novel Null Mutations in theEYSGene Are a Frequent Cause of Autosomal Recessive Retinitis Pigmentosa in the Israeli Population. , 2010, 51, 4387.		57
83	A Novel Nonsense Mutation inCEP290Induces Exon Skipping and Leads to a Relatively Mild Retinal Phenotype. , 2010, 51, 3646.		65
84	Homozygosity Mapping in Patients with Coneâ€“Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
85	Mutations in the EYS Gene Account for Approximately 5% of Autosomal Recessive Retinitis Pigmentosa and Cause a Fairly Homogeneous Phenotype. Ophthalmology, 2010, 117, 2026-2033.e7.	5.2	72
86	Novel CNGA3 and CNGB3 mutations in two Pakistani families with achromatopsia. Molecular Vision, 2010, 16, 774-81.	1.1	13
87	Missense mutations at homologous positions in the fourth and fifth laminin A G-like domains of eyes shut homolog cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2010, 16, 2753-9.	1.1	17
88	Audiometric and Vestibular Features in a Second Dutch DFNA20/26 Family with a Novel Mutation inACTG1. Annals of Otology, Rhinology and Laryngology, 2009, 118, 382-390.	1.1	20
89	Mutations in SPATA7 Cause Leber Congenital Amaurosis and Juvenile Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 380-387.	6.2	111
90	Homozygosity Mapping Reveals PDE6C Mutations in Patients with Early-Onset Cone Photoreceptor Disorders. American Journal of Human Genetics, 2009, 85, 240-247.	6.2	194

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91	A novel mutation in GRK1 causes Oguchi disease in a consanguineous Pakistani family. Molecular Vision, 2009, 15, 1788-93.	1.1	22
92	A homozygous p.Glu150Lys mutation in the opsin gene of two Pakistani families with autosomal recessive retinitis pigmentosa. Molecular Vision, 2009, 15, 2526-34.	1.1	15
93	Missense mutations inPOU4F3cause autosomal dominant hearing impairment DFNA15 and affect subcellular localization and DNA binding. Human Mutation, 2008, 29, 545-554.	2.5	62
94	Identification of a 2 Mb Human Ortholog of Drosophila eyes shut/spacemaker that Is Mutated in Patients with Retinitis Pigmentosa. American Journal of Human Genetics, 2008, 83, 594-603.	6.2	141
95	Mid-frequency DFNA8/12 hearing loss caused by a synonymous TECTA mutation that affects an exonic splice enhancer. European Journal of Human Genetics, 2008, 16, 1430-1436.	2.8	33
96	Mutations of LRTOMT, a fusion gene with alternative reading frames, cause nonsyndromic deafness in humans. Nature Genetics, 2008, 40, 1335-1340.	21.4	65
97	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	6.2	127
98	Phenotype Description of a Novel DFNA9/COCH Mutation, I109T. Annals of Otology, Rhinology and Laryngology, 2007, 116, 349-357.	1.1	29
99	Clinical Characteristics of a Dutch DFNA9 Family with a Novel COCH Mutation, G87W. Audiology and Neuro-Otology, 2007, 12, 77-84.	1.3	18
100	Involvement ofDFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. Human Mutation, 2007, 28, 718-723.	2.5	58
101	Biosynthesis and differential processing of two pools of amyloidâ€î² precursor protein in a physiologically inducible neuroendocrine cell. Journal of Neurochemistry, 2005, 94, 1015-1024.	3.9	12
102	Identification and expression of the first nonmammalian amyloid-beta precursor-like protein APLP2 in the amphibian Xenopus laevis. FEBS Journal, 2004, 271, 1906-1912.	0.2	9