Bart P Leroy

List of Publications by Year
in descending order

82 papers	4,255 citations	201674 27 h-index	123424 61 g-index
90 all docs	90 docs citations	90 times ranked	4899 citing authors

#	Article	IF	Citations
1	X-Linked Retinoschisis. Ophthalmology, 2022, 129, 191-202.	5.2	29
2	The Natural History of Leber Congenital Amaurosis and Coneâ€“Rod Dystrophy Associated with Variants in the GUCY2D Gene. Ophthalmology Retina, 2022, 6, 711-722.	2.4	8
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	The corneoscleral shape in Marfan syndrome. Acta Ophthalmologica, 2021, 99, 405-410.	1.1	8
5	Phenocopy of a heterozygous carrier of X-linked retinitis pigmentosa due to mosaicism for a RHO variant. Scientific Reports, 2021, 11, 117.	3.3	5
6	Clinical Perspective: Treating RPE65-Associated Retinal Dystrophy. Molecular Therapy, 2021, 29, 442-463.	8.2	92
7	LEBER CONGENITAL AMAUROSIS DUE TO CEP290 MUTATIONSâ€”SEVERE VISION IMPAIRMENT WITH A HIGH UNMET MEDICAL NEED. Retina, 2021, 41, 898-907.	1.7	24
8	CRB1-associated retinal dystrophies in a Belgian cohort: genetic characteristics and long-term clinical follow-up. British Journal of Ophthalmology, 2021, , bjophthalmol-2020-316781.	3.9	13
9	The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet Journal of Rare Diseases, 2021, 16, 142.	2.7	25
10	A Virtual Reality Orientation and Mobility Test for Inherited Retinal Degenerations: Testing a Proof-of-Concept After Gene Therapy. Clinical Ophthalmology, 2021, Volume 15, 939-952.	1.8	13
11	Mild Leber hereditary optic neuropathy (LHON) in a Western European family due to the rare Asian m.14502T>C variant in the MT-ND6 gene. Ophthalmic Genetics, 2021, 42, 440-445.	1.2	2
12	Longitudinal phenotypic study of late-onset retinal degeneration due to a founder variant c.562C>A p.(Pro188Thr) in theC1QTNF5gene. Ophthalmic Genetics, 2021, 42, 521-532.	1.2	10
13	Expanding the clinical spectrum and management of Traboulsi syndrome: report on two siblings homozygous for a novel pathogenic variant in ASPH. Ophthalmic Genetics, 2021, 42, 493-499.	1.2	5
14	A qualitative study among patients with an inherited retinal disease on the meaning of genomic unsolicited findings. Scientific Reports, 2021, 11, 15834.	3.3	5
15	US Health Resource Utilization and Cost Burden Associated with Choroideremia. Clinical Ophthalmology, 2021, Volume 15, 3459-3465.	1.8	4
16	The Phenotypic Spectrum of Patients with PHARC Syndrome Due to Variants in ABHD12: An Ophthalmic Perspective. Genes, 2021, 12, 1404.	2.4	7
17	Durability of Voretigene Neparvovec for Biallelic RPE65-Mediated Inherited Retinal Disease. Ophthalmology, 2021, 128, 1460-1468.	5.2	82
18	Broad locations of antigenic regions for anti-TRPM1 autoantibodies in paraneoplastic retinopathy with retinal ON bipolar cell dysfunction. Experimental Eye Research, 2021, 212, 108770.	2.6	3

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19	CLINICAL CHARACTERISTICS AND NATURAL HISTORY OF RHO-ASSOCIATED RETINITIS PIGMENTOSA. Retina, 2021, 41, 213-223.	1.7	18
20	89â€...Five-year update for the Phase III voretigene neparvovec study in biallelic RPE65 mutationâ€“associated inherited retinal disease. , 2021, , .		1
21	ISOLATED MACULOPATHY AND MODERATE RODâ€“CONE DYSTROPHY REPRESENT THE MILDER END OF THE RDH12-RELATED RETINAL DYSTROPHY SPECTRUM. Retina, 2021, 41, 1346-1355.	1.7	4
22	Comparative Natural History of Visual Function From Patients With Biallelic Variants inBBS1andBBS10. , 2021, 62, 26.		11
23	Genetic testing and diagnosis of inherited retinal diseases. Orphanet Journal of Rare Diseases, 2021, 16, 514.	2.7	25
24	Functional characterization of novel MFSD8 pathogenic variants anticipates neurological involvement in juvenile isolated maculopathy. Clinical Genetics, 2020, 97, 426-436.	2.0	11
25	Advancing Clinical Trials for Inherited Retinal Diseases: Recommendations from the Second Monaciano Symposium. Translational Vision Science and Technology, 2020, 9, 2.	2.2	56
26	Vitreous Hemorrhage as Presenting Sign of Retinal Arteriovenous Malformation. Case Reports in Ophthalmological Medicine, 2020, 2020, 1-4.	0.5	1
27	Hydroxychloroquine hitting the headlinesâ€”retinal considerations. Eye, 2020, 34, 1158-1160.	2.1	7
28	The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in MFRP and PRSS56. Scientific Reports, 2020, 10, 1289.	3.3	24
29	Functional characterization of the first missense variant in CEP78 , a founder allele associated with coneâ€rod dystrophy, hearing loss, and reduced male fertility. Human Mutation, 2020, 41, 998-1011.	2.5	15
30	<scp>VEGFA</scp> variants as prognostic markers for the retinopathy in pseudoxanthoma elasticum. Clinical Genetics, 2020, 98, 74-79.	2.0	8
31	Long-Term Follow-Up of Retinal Degenerations Associated WithLRATMutations and Their Comparability to Phenotypes Associated WithRPE65Mutations. Translational Vision Science and Technology, 2019, 8, 24.	2.2	14
32	Efficacy, Safety, and Durability of Voretigene Neparvovec-rzyl in RPE65 Mutationâ€“Associated Inherited Retinal Dystrophy. Ophthalmology, 2019, 126, 1273-1285.	5.2	239
33	Mitochondrial single-stranded DNA binding protein novel de novo SSBP1 mutation in a child with single large-scale mtDNA deletion (SLSMD) clinically manifesting as Pearson, Kearns-Sayre, and Leigh syndromes. PLoS ONE, 2019, 14, e0221829.	2.5	32
34	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
35	The Nâ€terminal p.(Ser38Cys) TIMP3 mutation underlying Sorsby fundus dystrophy is a founder mutation disrupting an intramolecular disulfide bond. Human Mutation, 2019, 40, 539-551.	2.5	10
36	Where are the missing gene defects in inherited retinal disorders? Intronic and synonymous variants contribute at least to 4% of CACNA1F â€mediated inherited retinal disorders. Human Mutation, 2019, 40, 765-787.	2.5	24

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37	Effect of an intravitreal antisense oligonucleotide on vision in Leber congenital amaurosis due to a photoreceptor cilium defect. Nature Medicine, 2019, 25, 225-228.	30.7	177
38	Ocular involvement in systemic sclerosis: A systematic literature review, it's not all scleroderma that meets the eye. Seminars in Arthritis and Rheumatism, 2019, 49, 119-125.	3.4	26
39	Abetalipoproteinemia From Previously Unreported Gene Mutations. Annals of Internal Medicine, 2019, 170, 211.	3.9	7
40	Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. Genetics in Medicine, 2019, 21, 1319-1329.	2.4	15
41	The Natural History of Inherited Retinal Dystrophy Due to Biallelic Mutations in the RPE65 Gene. American Journal of Ophthalmology, 2019, 199, 58-70.	3.3	77
42	Mapping the genomic landscape of inherited retinal disease genes prioritizes genes prone to coding and noncoding copy-number variations. Genetics in Medicine, 2018, 20, 202-213.	2.4	47
43	Analysis of <scp>KERA</scp> in four families with cornea plana identifies two novel mutations. Acta Ophthalmologica, 2018, 96, e87-e91.	1.1	4
44	Pathogenic variants in the ABCC6 gene are associated with an increased risk for ischemic stroke. Brain Pathology, 2018, 28, 822-831.	4.1	28
45	Clinical Characterization of 66 Patients With Congenital Retinal Disease Due to the Deep-Intronic c.2991+1655A>G Mutation inCEP290. , 2018, 59, 4384.		21
46	Mutations in the gene PDE6C encoding the catalytic subunit of the cone photoreceptor phosphodiesterase in patients with achromatopsia. Human Mutation, 2018, 39, 1366-1371.	2.5	18
47	CUGC for congenital primary aphakia. European Journal of Human Genetics, 2018, 26, 1234-1237.	2.8	4
48	The Spectrum of Structural and Functional Abnormalities in Female Carriers of Pathogenic Variants in theRPGRGene. , 2018, 59, 4123.		41
49	Diplopia as presenting sign of Turcot syndrome. International Ophthalmology, 2017, 37, 275-278.	1.4	2
50	Isolated maculopathy associated with biallelic CRB1 mutations. Ophthalmic Genetics, 2017, 38, 190-193.	1.2	16
51	Unilateral cancer-associated retinopathy: diagnosis, serology and treatment. Documenta Ophthalmologica, 2017, 135, 233-240.	2.2	25
52	Efficacy and safety of voretigene neparvovec (AAV2-hRPE65v2) in patients with RPE65 -mediated inherited retinal dystrophy: a randomised, controlled, open-label, phase 3 trial. Lancet, The, 2017, 390, 849-860.	13.7	1,250
53	Early-onset primary antibody deficiency resembling common variable immunodeficiency challenges the diagnosis of Wiedeman-Steiner and Roifman syndromes. Scientific Reports, 2017, 7, 3702.	3.3	30
54	Copy-number variation is an important contributor to the genetic causality of inherited retinal degenerations. Genetics in Medicine, 2017, 19, 643-651.	2.4	51

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55	arrEYE: a customized platform for high-resolution copy number analysis of coding and noncoding regions of known and candidate retinal dystrophy genes and retinal noncoding RNAs. Genetics in Medicine, 2017, 19, 457-466.	2.4	39
56	Mutations in Splicing Factor Genes Are a Major Cause of Autosomal Dominant Retinitis Pigmentosa in Belgian Families. PLoS ONE, 2017, 12, e0170038.	2.5	47
57	The importance of genetic testing as demonstrated by two cases of -associated retinal generation misdiagnosed as LCA. Molecular Vision, 2017, 23, 695-706.	1.1	13
58	Safety and durability of effect of contralateral-eye administration of AAV2 gene therapy in patients with childhood-onset blindness caused by RPE65 mutations: a follow-on phase 1 trial. Lancet, The, 2016, 388, 661-672.	13.7	377
59	Autosomal recessive retinitis pigmentosa with homozygous rhodopsin mutation E150K and non-coding cis-regulatory variants in CRX-binding regions of SAMD7. Scientific Reports, 2016, 6, 21307.	3.3	16
60	Detailed functional and structural phenotype of Bietti crystalline dystrophy associated with mutations in CYP4V2 complicated by choroidal neovascularization. Ophthalmic Genetics, 2016, 37, 445-452.	1.2	34
61	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. American Journal of Human Genetics, 2016, 99, 470-480.	6.2	39
62	De novo intrachromosomal gene conversion from OPN1MW to OPN1LW in the male germline results in Blue Cone Monochromacy. Scientific Reports, 2016, 6, 28253.	3.3	28
63	Do not turn a blind eye to alkyl nitrite (poppers)!. Acta Ophthalmologica, 2016, 94, e82-e83.	1.1	13
64	Visual Prognosis in USH2A-Associated Retinitis Pigmentosa Is Worse for Patients with Usher Syndrome Type IIa Than for Those with Nonsyndromic Retinitis Pigmentosa. Ophthalmology, 2016, 123, 1151-1160.	5.2	76
65	Mutations in CTNNA1 cause butterfly-shaped pigment dystrophy and perturbed retinal pigment epithelium integrity. Nature Genetics, 2016, 48, 144-151.	21.4	50
66	A Nonsense Mutation inFAM161AIs a Recurrent Founder Allele in Dutch and Belgian Individuals With Autosomal Recessive Retinitis Pigmentosa. , 2015, 56, 7418.		9
67	Cobalamin C Deficiency Shows a Rapidly Progressing Maculopathy With Severe Photoreceptor and Ganglion Cell Loss. , 2015, 56, 7875.		30
68	Novel FRMD7 Mutations and Genomic Rearrangement Expand the Molecular Pathogenesis of X-Linked Idiopathic Infantile Nystagmus. Investigative Ophthalmology and Visual Science, 2015, 56, 1701-1710.	3.3	25
69	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
70	Colour Vision in Stargardt Disease. Ophthalmic Research, 2015, 54, 181-194.	1.9	11
71	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	6.2	42
72	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

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73	Hidden Genetic Variation in LCA9â€Associated Congenital Blindness Explained by 5â€²UTR Mutations and Copyâ€Number Variations of NMNAT1. Human Mutation, 2015, 36, 1188-1196.	2.5	30
74	Mutations in IFT172 cause isolated retinal degeneration and Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2015, 24, 230-242.	2.9	136
75	Abnormal retinal development associated with FRMD7 mutations. Human Molecular Genetics, 2014, 23, 4086-4093.	2.9	70
76	SLC24A5 Mutations Are Associated with Non-Syndromic Oculocutaneous Albinism. Journal of Investigative Dermatology, 2014, 134, 568-571.	0.7	36
77	Identity-by-descentâ€“guided mutation analysis and exome sequencing in consanguineous families reveals unusual clinical and molecular findings in retinal dystrophy. Genetics in Medicine, 2014, 16, 671-680.	2.4	53
78	Spectrum of Cav1.4 dysfunction in congenital stationary night blindness type 2. Biochimica Et Biophysica Acta - Biomembranes, 2014, 1838, 2053-2065.	2.6	26
79	Genetic screening of LCA in Belgium: predominance of CEP290 and identification of potential modifier alleles in AHI1 of CEP290-related phenotypes. Human Mutation, 2010, 31, E1709-E1766.	2.5	127
80	Ptosis as an associated finding in maternally inherited diabetes and deafness. Ophthalmic Genetics, 2010, 31, 240-243.	1.2	7
81	Cancer-associated retinopathy (CAR) with electronegative ERG: a case report. Documenta Ophthalmologica, 2008, 116, 49-55.	2.2	23
82	Clinical Features & Retinal Function In Patients With Adult Refsum Syndrome. Advances in Experimental Medicine and Biology, 2004, 544, 57-58.	1.6	6