Review and update on the molecular basis of Leber congenital amaurosis

DOI: 10.12998/wjcc.v3.i2.112
World Journal of Clinical Cases, 2015, 3, 112-24.

#	Paper	IF	Citations
82	Using patient-specific induced pluripotent stem cells to interrogate the pathogenicity of a novel retinal pigment epithelium-specific 65 kDa cryptic splice site mutation and confirm eligibility for enrollment into a clinical gene augmentation trial. *Translational Research*, **2015**, 166, 740-749.e1	11	22
81	Investor Outlook: Focus on Upcoming LCA2 Gene Therapy Phase III Results. *Human Gene Therapy Clinical Development*, **2015**, 26, 144-9	3.2	15
80	Non-viral therapeutic approaches to ocular diseases: An overview and future directions. *Journal of Controlled Release*, **2015**, 219, 471-487	11.7	34
79	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases. *Journal of Human Genetics*, **2016**, 61, 951-958	4.3	6
78	Hereditary Retinal Dystrophy. *Handbook of Experimental Pharmacology*, **2017**, 242, 337-367	3.2	22
77	Identification and Correction of Mechanisms Underlying Inherited Blindness in Human iPSC-Derived Optic Cups. *Cell Stem Cell*, **2016**, 18, 769-781	18	193
76	Phenotypic features of CRB1-associated early-onset severe retinal dystrophy and the different molecular approaches to identifying the disease-causing variants. *Graefeks Archive for Clinical and Experimental Ophthalmology*, **2016**, 254, 1833-9	3.8	11
75	Clinical and genetic characteristics of Leber congenital amaurosis with novel mutations in known genes based on a Chinese eastern coast Han population. *Graefeks Archive for Clinical and Experimental Ophthalmology*, **2016**, 254, 2227-2238	3.8	20
74	Gene and cell-based therapies for inherited retinal disorders: An update. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2016**, 172, 349-366	3.1	50
73	Intravenous delivery of adeno-associated virus 9-encoded IGF-1Ea propeptide improves post-infarct cardiac remodelling. *Npj Regenerative Medicine*, **2016**, 1, 16001	15.8	5
72	Neonatal disease environment limits the efficacy of retinal transplantation in the LCA8 mouse model. *BMC Ophthalmology*, **2016**, 16, 193	2.3	0
71	Genome-editing Technologies for Gene and Cell Therapy. *Molecular Therapy*, **2016**, 24, 430-46	11.7	413
70	Ion Channels in the Eye: Involvement in Ocular Pathologies. *Advances in Protein Chemistry and Structural Biology*, **2016**, 104, 157-231	5.3	15
69	A Dominant Mutation in Rpe65, D477G, Delays Dark Adaptation and Disturbs the Visual Cycle in the Mutant Knock-In Mice. *American Journal of Pathology*, **2017**, 187, 517-527	5.8	15
68	Defining Outcomes for Clinical Trials of Leber Congenital Amaurosis Caused by GUCY2D Mutations. *American Journal of Ophthalmology*, **2017**, 177, 44-57	4.9	21
67	Phenotype-genotype correlation with Sanger sequencing identified retinol dehydrogenase 12 (RDH12) compound heterozygous variants in a Chinese family with Leber congenital amaurosis. *Journal of Zhejiang University: Science B*, **2017**, 18, 421-429	4.5	7
66	Homozygosity Mapping for Autosomal Recessive Ocular Diseases. *Essentials in Ophthalmology*, **2017**, 449-456	0.2	

65	Induced Pluripotent Stem Cell-Derived Autologous Cell Therapy for Age-Related Macular Degeneration. **2017**, 33-44		
64	Mutations in the PCYT1A gene are responsible for isolated forms of retinal dystrophy. *European Journal of Human Genetics*, **2017**, 25, 651-655	5.3	14
63	The clinical implications of molecular monitoring and analyses of inherited retinal diseases. *Expert Review of Molecular Diagnostics*, **2017**, 17, 1009-1021	3.8	5
62	Chlamydomonas: Cilia and Ciliopathies. *Microbiology Monographs*, **2017**, 73-97	0.8	
61	Clinical and Genetic Evaluation of a Cohort of Pediatric Patients with Severe Inherited Retinal Dystrophies. *Genes*, **2017**, 8,	4.2	16
60	Molecular Screening of 43 Brazilian Families Diagnosed with Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy. *Genes*, **2017**, 8,	4.2	14
59	Prescreening whole exome sequencing results from patients with retinal degeneration for variants in genes associated with retinal degeneration. *Clinical Ophthalmology*, **2018**, 12, 49-63	2.5	15
58	Insights into the pathogenesis of dominant retinitis pigmentosa associated with a D477G mutation in RPE65. *Human Molecular Genetics*, **2018**, 27, 2225-2243	5.6	21
57	Leberև Congenital Amaurosis and Gene Therapy. *Indian Journal of Pediatrics*, **2018**, 85, 237-242	3	7
56	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing. *Clinical Genetics*, **2018**, 93, 329-339	4	12
55	Safety and Long-Term Efficacy of AAV4 Gene Therapy in Patients with RPE65 Leber Congenital Amaurosis. *Molecular Therapy*, **2018**, 26, 256-268	11.7	63
54	Children with blindness - major causes, developmental outcomes and implications for habilitation and educational support: a two-decade, Swedish population-based study. *Acta Ophthalmologica*, **2018**, 96, 295-300	3.7	24
53	Identification of a novel RPGRIP1 mutation in an Iranian family with leber congenital amaurosis by exome sequencing. *Journal of Cellular and Molecular Medicine*, **2018**, 22, 1733-1742	5.6	15
52	Mutation in the Zebrafish cct2 Gene Leads to Abnormalities of Cell Cycle and Cell Death in the Retina: A Model of CCT2-Related Leber Congenital Amaurosis. **2018**, 59, 995-1004		13
51	Review of Ocular Manifestations of Joubert Syndrome. *Genes*, **2018**, 9,	4.2	29
50	RDH12 Mutations Cause a Severe Retinal Degeneration With Relatively Spared Rod Function. **2018**, 59, 5225-5236		30
49	Roles of Nmnat1 in the survival of retinal progenitors through the regulation of pro-apoptotic gene expression via histone acetylation. *Cell Death and Disease*, **2018**, 9, 891	9.8	13
48	Preimplantation genetic diagnosis as a strategy to prevent having a child born with an heritable eye disease. *Ophthalmic Genetics*, **2018**, 39, 450-456	1.2	15

47	Rhythmic Regulation of Photoreceptor and RPE Genes Important for Vision and Genetically Associated With Severe Retinal Diseases. **2018**, 59, 3789-3799		8
46	Translation of CRISPR Genome Surgery to the Bedside for Retinal Diseases. *Frontiers in Cell and Developmental Biology*, **2018**, 6, 46	5.7	15
45	Leber congenital amaurosis: Current genetic basis, scope for genetic testing and personalized medicine. *Experimental Eye Research*, **2019**, 189, 107834	3.7	14
44	Pluripotent Stem Cells to Model Degenerative Retinal Diseases: The RPE Perspective. *Advances in Experimental Medicine and Biology*, **2019**, 1186, 1-31	3.6	5
43	Targeted deletion of Crb1/Crb2 in the optic vesicle models key features of leber congenital amaurosis 8. *Developmental Biology*, **2019**, 453, 141-154	3.1	2
42	is required for the integrity of the photoreceptor layer in the zebrafish retina. *Biology Open*, **2019**, 8,	2.2	2
41	Ligand Binding Mechanisms in Human Cone Visual Pigments. *Trends in Biochemical Sciences*, **2019**, 44, 629-639	10.3	2
40	Retina Genes in Chinese. *Essentials in Ophthalmology*, **2019**, 177-190	0.2	
39	Gene therapy and the adeno-associated virus in the treatment of genetic and acquired ophthalmic diseases in humans: Trials, future directions and safety considerations. *Clinical and Experimental Ophthalmology*, **2019**, 47, 521-536	2.4	24
38	A Novel Nonsense Mutation: Case Report of Three Siblings Diagnosed with Leber Congenital Amaurosis. *Fetal and Pediatric Pathology*, **2020**, 39, 251-258	1.7	0
37	Possible dual contribution of a novel GUCY2D mutation in the development of retinal degeneration in a consanguineous population. *European Journal of Medical Genetics*, **2020**, 63, 103750	2.6	
36	Genetic and clinical findings in a Chinese cohort with Leber congenital amaurosis and early onset severe retinal dystrophy. *British Journal of Ophthalmology*, **2020**, 104, 932-937	5.5	15
35	Clinical and Genetic Analysis of a European Cohort with Pericentral Retinitis Pigmentosa. *International Journal of Molecular Sciences*, **2019**, 21,	6.3	13
34	Outer retinal degeneration in two closely related Goeldi's monkeys (Callimico goeldii). *Veterinary Ophthalmology*, **2020**, 23, 394-401	1.4	
33	Clinical and molecular findings in a cohort of 152 Brazilian severe early onset inherited retinal dystrophy patients. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2020**, 184, 728-752	3.1	9
32	The Aryl Hydrocarbon Receptor: A Mediator and Potential Therapeutic Target for Ocular and Non-Ocular Neurodegenerative Diseases. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	6
31	Transient pupillary light reflex in CEP290- or NPHP5-associated Leber congenital amaurosis: Latency as a potential outcome measure of cone function. *Vision Research*, **2020**, 168, 53-63	2.1	9
30	Hereditary Chorioretinal Disorders. *Retina Atlas*, **2020**,	0	

29	The pathogenicity of novel GUCY2D mutations in Leber congenital amaurosis 1 assessed by HPLC-MS/MS. *PLoS ONE*, **2020**, 15, e0231115	3.7	0
28	Zebrafish Models of Photoreceptor Dysfunction and Degeneration. *Biomolecules*, **2021**, 11,	5.9	2
27	-associated retinal dystrophies in a Belgian cohort: genetic characteristics and long-term clinical follow-up. *British Journal of Ophthalmology*, **2021**,	5.5	6
26	Leber congenital amaurosis/early-onset severe retinal dystrophy: current management and clinical trials. *British Journal of Ophthalmology*, **2021**,	5.5	7
25	An Update on Gene Therapy for Inherited Retinal Dystrophy: Experience in Leber Congenital Amaurosis Clinical Trials. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	10
24	RNA therapeutics in ophthalmology - translation to clinical trials. *Experimental Eye Research*, **2021**, 205, 108482	3.7	9
23	Molecular genetics with clinical characteristics of Leber congenital amaurosis in the Han population of western China. *Ophthalmic Genetics*, **2021**, 42, 392-401	1.2	0
22	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India. *Eye and Vision (London, England)*, **2021**, 8, 20	4.9	1
21	The Timecourses of Functional, Morphological, and Molecular Changes Triggered by Light Exposure in Sprague-Dawley Rat Retinas. *Cells*, **2021**, 10,	7.9	7
20	Spectrum of Disease Severity in Nonsyndromic Patients With Mutations in the CEP290 Gene: A Multicentric Longitudinal Study. **2021**, 62, 1		0
19	Leberև Congenital Amaurosis: Current Concepts of Genotype-Phenotype Correlations. *Genes*, **2021**, 12,	4.2	2
18	Genome editing in large animal models. *Molecular Therapy*, **2021**, 29, 3140-3152	11.7	2
17	Ligand-based rational design, synthesis and evaluation of novel potential chemical chaperones for opsin. *European Journal of Medicinal Chemistry*, **2021**, 226, 113841	6.8	0
16	Retinitis Pigmentosa. *Retina Atlas*, **2020**, 1-43	0	
15	Intravitreal Sepofarsen for Leber Congenital Amaurosis Type 10 (LCA10). *SSRN Electronic Journal*,	1	
14	CRB1-Related Leber Congenital Amaurosis: Reporting Novel Pathogenic Variants and a Brief Review on Mutations Spectrum. *Iranian Biomedical Journal*, **2019**, 23, 362-8	2	
13	Copy number variations and multiallelic variants in Korean patients with Leber congenital amaurosis. *Molecular Vision*, **2020**, 26, 26-35	2.3	8
12	Modeling inherited retinal dystrophies using induced pluripotent stem cells. **2022**, 157-184		

11	The Genetic Landscape of Inherited Retinal Diseases in a Mexican Cohort: Genes, Mutations and Phenotypes. *Genes*, **2021**, 12,	4.2	1
10	Clinical Features and Natural History in a Cohort of Chinese Patients with RPE65-Associated Inherited Retinal Dystrophy. *Journal of Clinical Medicine*, **2021**, 10,	5.1	
9	Review of gene therapies for age-related macular degeneration.. *Eye*, **2022**,	4.4	2
8	Exome sequencing identifies a novel GUCY2D mutation in an Iranian family with Leber congenital amaurosis-1: a case report. *Egyptian Journal of Medical Human Genetics*, **2022**, 23,	2	
7	Intravitreal antisense oligonucleotide sepofarsen in Leber congenital amaurosis type 10: a phase 1b/2 trial.. *Nature Medicine*, **2022**,	50.5	5
6	TUBB4B gene mutation in Leber phenotype of congenital amaurosis syndrome associated with early-onset deafness.. *European Journal of Medical Genetics*, **2022**, 65, 104471	2.6	0
5	Mobility test to assess functional vision in dark-adapted patients with Leber congenital amaurosis. *BMC Ophthalmology*, **2022**, 22,	2.3	0
4	Publication Trends of Leber Congenital Amaurosis Researches: A Bibliometric Study during 2002-2021.		0
3	A novel missense ALMS1 variant causes aberrant splicing identified in a cohort of patients with Alstrm syndrome. 13,		0
2	Leber Congenital Amaurosis: Leading Cause of Inherited Blindness in Children. **2023**, 19, 104560		0
1	Principais achados em um paciente com diagnstico de Amaurose Congnita de Leber. **2023**, 82,		0