

Ongoing clinical trials of gene- and gene-specific therapy for Inherited Retinal Disorders

Inherited Retinal Disease (<i>causative gene mutation</i>)	Intervention(s)	Study Sponsor	Country	Clinical Phase	Study name / ClinicalTrials.gov Identifier	Recruitment status	Estimated primary study completion date
Leber Congenital Amaurosis (<i>RPE65</i>)	rAAV2-CBSB-hRPE65	University of Pennsylvania/ National Eye Institute (NEI)	USA	Phase 1	NCT00481546	Active (not recruiting)	June 2026
Leber Congenital Amaurosis (<i>RPE65</i>)	AAV2/5-OPTIRPE65	MeiraGTx UK II Ltd	UK	Phase 1/2	NCT02781480	Recruiting	October 2018
Leber Congenital Amaurosis (<i>RPE65</i>)	AAV2/5-OPTIRPE65	MeiraGTx UK II Ltd	UK	Phase 1/2	Long-term follow-up of above study NCT02946879	Recruiting	April 2023
Leber Congenital Amaurosis (<i>RPE65</i>)	Voretigene neparvovec-rzyl	Spark Therapeutics	USA	Phase 1	NCT00516477	Active, not recruiting	July 2024
Leber Congenital Amaurosis (<i>RPE65</i>)	Voretigene neparvovec-rzyl	Spark Therapeutics	USA	Phase 3	NCT00999609	Active, not recruiting	July 2015
Leber Congenital Amaurosis (<i>RPE65</i>)	Voretigene neparvovec-rzyl	Spark Therapeutics	Not specified	5-year registry study	NCT03597399	Not yet recruiting	August 2028
Leber Congenital Amaurosis (<i>CEP290</i>)	QR-110 RNA-based therapy	ProQR Therapeutics	USA Belgium	Phase 1/2	NCT03140969	Active, not recruiting	December 2019

X-linked juvenile retinoschisis (RS1)	RS1 AAV Vector	National Eye Institute (NEI)	USA	Phase 1/2	NCT02317887	Recruiting	July 2021
X-linked juvenile retinoschisis (RS1)	rAAV2tYF-CB-hRS1	Applied Genetic Technologies Corp	USA	Phase 1/2	NCT02416622	Recruiting	October 2018
X-linked Retinitis Pigmentosa (RPGR)	AAV-RPGR	Nightstar Therapeutics	UK	Phase 1/2	XIRIUS NCT03116113	Recruiting	February 2019
X-linked Retinitis Pigmentosa (RPGR)	AAV-RPGR	MeiraGTx UK II Ltd	UK	Phase 1/2	NCT03252847	Recruiting	November 2020
X-linked Retinitis Pigmentosa (RPGR-ORF15)	rAAV2tYF-GRK1-RPGR	Applied Genetic Technologies Corp	USA	Phase 1/2	NCT03316560	Recruiting	January 2022
Retinitis Pigmentosa (RLBP1)	CPK850	Novartis Pharmaceuticals	Sweden	Phase 1/2	NCT03374657	Recruiting	September 2025
Retinitis Pigmentosa (PDE6A)	N/A – observational study	STZ eyetrial: clinical characterization preparation study for Phase 1/2 gene therapy trial	Germany	N/A	NCT02759952	Enrolling by invitation	December 2020
Retinitis Pigmentosa (PDE6B)	AAV2/5-hPDE6B	Horama S.A.	France	Phase 1/2	NCT03328130	Recruiting	June 2020
Retinitis Pigmentosa	RST-001	Allergan	USA	Phase 1/2	NCT02556736	Recruiting	August 2019

Retinitis Pigmentosa	GS030-DP (gene therapy) & Medical device: GS030-MD	GenSight Biologics	UK	Phase 1/2	PIONEER NCT03326336	Not yet recruiting	June 2020
Choroideremia (CHM)	AAV2-REP1	Nightstar Therapeutics	USA	Phase 2	GEMINI NCT03507686	Recruiting	March 2020
Choroideremia (CHM)	AAV2-REP1	Nightstar Therapeutics	USA Canada Finland Germany Netherland UK	Phase 3	STAR NCT03496012	Recruiting	March 2020
Choroideremia (CHM)	rAAV2.REP1	STZ eyetrial & University Hospital Tübingen	Germany	Phase 2	THOR NCT02671539	Active, not recruiting	March 2018
Choroideremia (CHM)	AAV-mediated REP1 gene replacement	University of Oxford	UK	Phase 2	REGENERATE NCT02407678	Recruiting	August 2019
Choroideremia (CHM)	AAV2-hCHM	Spark Therapeutics	United States	Phase 1/2	NCT02341807	Active, not recruiting	January 2019
Achromatopsia (CNGB3)	AAV-CNGB3	MeiraGTx UK II Ltd	UK	Phase 1/2	NCT03001310	Recruiting	February 2019
Achromatopsia (CNGB3)	rAAV2tYF-PR1.7-hCNGB3	Applied Genetic Technologies Corp. & National Eye Institute (NEI)	USA	Phase 1/2	NCT02599922	Recruiting	December 2018
Achromatopsia (CNGA3)	AGTC-402	Applied Genetic Technologies Corp	USA, Israel	Phase 1/2	NCT02935517	Recruiting	June 2019

Achromatopsia (<i>CNGA3</i>)	rAAV.hCNGA3	STZ eyetrial University Hospital Tübingen	Germany	Phase 1/2	NCT02610582	Active, not recruiting	November 2017
Usher syndrome type 1B (<i>MYO7A</i>)	UshStat	Sanofi	US and France	Phase 1/2	NCT01505062	Recruiting	January 2021
Stargardt disease (<i>ABCA4</i>)	SAR422459	Sanofi	US and France	Phase 1/2	NCT01367444	Recruiting	November 2019
Leber Hereditary Optic Neuropathy (<i>G11778A</i>)	rAAV2-ND4	Huazhong University of Science and Technology	China	Phase 2/3	NCT03153293	Active, not recruiting	June 2019
Leber's Hereditary Optic Neuropathy (<i>G11778A</i>)	scAAV2- P1ND4v2	National Eye Institute (NEI)	USA	Phase 1	NCT02161380	Recruiting	March 2019
Leber Hereditary Optic Neuropathy (<i>ND4</i>)	GS010 (rAAV2/2-ND4)	GenSight Biologics	France	Phase 1/2	NCT02064569	Active, not recruiting	June 2020
Leber Hereditary Optic Neuropathy (<i>ND4</i>)	GS010	GenSight Biologics	USA France Germany Italy UK	Phase 3	RESCUE NCT02652767	Active, not recruiting	September 2018
Leber Hereditary Optic Neuropathy (<i>ND4</i>)	GS010	GenSight Biologics	USA France Germany Italy UK	Phase 3	REVERSE NCT02652780	Active, not recruiting	January 2018
Leber's Hereditary Optic Neuropathy (<i>ND4</i>)	GS010	GenSight Biologics	USA Belgium Taiwan	Phase 3	REFLECT NCT03293524	Recruiting	June 2020

Leber's Hereditary Optic Neuropathy (ND4)	GS010	GenSight Biologics	USA France Germany Italy UK		Long-term follow-up of RESCUE and REVERSE Phase 3 trials NCT03406104	Recruiting	July 2022
Leber's Hereditary Optic Neuropathy (ND4)	GS010	GenSight Biologics	Not specified	N/A	Expanded access program NCT03672968	Recruiting	N/A

Ongoing clinical trials of cell therapy for Inherited Retinal Disorders

Inherited Retinal Disease	Intervention(s)	Study Sponsor	Country	Clinical Phase	ClinicalTrials.gov Identifier	Recruitment status	Estimated primary study completion date
Stargardt Disease	MA09-hRPE cells	Astellas Institute for Regenerative Medicine	USA	Not specified	NCT02445612	Active, not recruiting	December 2019
Stargardt Disease	hESC-RPE cells	Astellas Institute for Regenerative Medicine	UK	Long-term follow-up of completed Phase 1/2 study	NCT02941991	Active, not recruiting	December 2019
Stargardt Disease	hESC-RPE cells	Southwest Hospital, China	China	Phase 1/2	NCT02749734	Active, not recruiting	December 2018
Stargardt Disease	hESC RPE cells	Federal University of São Paulo	Brazil	Phase 1/2	NCT02903576	Recruiting	June 2018
<ul style="list-style-type: none"> • Leber Congenital Amaurosis • Retinitis Pigmentosa 	HuRPE cells	Eyeecure Therapeutics Inc.	China	Phase 1	NCT03566147	Recruiting	December 2019

Retinitis Pigmentosa	Human retinal progenitor cells	jCyte, Inc	USA	Phase 2	NCT03073733	Active, not recruiting	August 2019
Retinitis Pigmentosa	hRPC cells	ReNeuron Limited	USA	Phase 1/2	NCT02464436	Recruiting	July 2018

Ongoing clinical trials of other treatments / interventions for Inherited Retinal Disorders

Inherited Retinal Disease	Intervention(s)	Study Sponsor	Country	Clinical Phase	ClinicalTrials.gov Identifier	Recruitment status	Estimated primary study completion date
Stargardt Disease	Dietary supplement of Omega-3 Fatty Acids	Ophthalmos Research and Education Institute	Not specified	Not specified	Macular Degeneration Omega-3 Study (MADEOS) NCT03297515	Not yet recruiting	December 2018
Stargardt Disease (ABCA4)	ALK-001 (C20-D3-retinyl acetate)	Alkeus Pharmaceuticals	USA	Phase 2	NCT02402660	Recruiting	July 2019
Stargardt disease (ABCA4)	Zimura (complement factor C5 inhibitor)	Ophthotech Corporation	Not specified	Phase 2	NCT03364153	Recruiting	September 2020
Stargardt Disease	Saffron supplementation	Catholic University of the Sacred Heart	Italy	Phase 1/2	NCT01278277	Active, not recruiting	November 2017
Leber Hereditary Optic Neuropathy	Elamipretide (MTP-131) 1% topical ophthalmic solution	Stealth BioTherapeutics Inc.	USA	Phase 2	NCT02693119	Active, not recruiting	May 2018
Leber Hereditary Optic Neuropathy	Idebenone	Santhera Pharmaceuticals	Austria France Germany Netherlands	Post-authorisation safety study	PAROS NCT02771379	Recruiting	February 2021
Retinitis Pigmentosa	N-Acetyl Cysteine (NAC)	Johns Hopkins University	USA	Phase 1	FIGHT-RP1 Study NCT03063021	Active, not recruiting	February 2019

<ul style="list-style-type: none"> • Retinitis Pigmentosa • Usher Syndrome Type 2 • Usher Syndrome Type 3 	NT-501 (Ciliary Neurotrophic Factor-Releasing Encapsulated Cell Implant)	Neurotech Pharmaceuticals	USA	Phase 2	NCT01530659	Active, not recruiting	August 2019
Retinitis Pigmentosa	Dietary supplement: Alga Dunaliella Bardaw	Sheba Medical Center	Israel	Phase 2/3	NCT01680510	Recruiting	December 2018
Retinitis Pigmentosa	Argus II Retinal Prosthesis System	Second Sight Medical Products	France	Not applicable	Post-Market Study of the Argus® II Retinal Prosthesis System NCT02303288	Active, not recruiting	November 2018
Retinitis Pigmentosa	Argus II Retinal Prosthesis	Second Sight Medical Products	Germany	Not applicable	Better Vision RP Study NCT03418116	Recruiting	November 2021
Retinitis Pigmentosa	Argus II eyeglasses /ORCAM Device	Mayo Clinic, Second Sight Medical Products	USA	Not applicable	Argus II/ORCAM Device study NCT03248388	Recruiting	June 2018
Retinitis Pigmentosa	Argus® II Retinal Prosthesis System	Second Sight Medical Products	USA	Not applicable	NCT01860092	Recruiting	August 2023
Retinitis Pigmentosa	Argus® II Retinal Prosthesis System	Second Sight Medical Products	Germany Italy	Post-market surveillance study	NCT01490827	Recruiting	December 2018
Retinitis Pigmentosa	Computer assisted rehabilitation environment	The Cleveland Clinic	USA	Not applicable	NCT03444961	Recruiting	July 2018

	(CAREN) system for Argus recipients						
Retinitis Pigmentosa	RETINA IMPLANT Alpha	Retina Implant AG	Germany	Not applicable	Post-marketing surveillance study NCT02588430	Enrolling by invitation	December 2020
<ul style="list-style-type: none"> • Retinitis Pigmentosa • Choroideremia 	RETINA IMPLANT Alpha AMS	Retina Implant AG	France	Not applicable	NCT03561922	Not yet recruiting	October 2021
Retinitis Pigmentosa	RETINA IMPLANT Alpha AMS	Wills Eye/Retina Implant AG	USA	Not applicable	NCT03629899	Recruiting	July 2025
<ul style="list-style-type: none"> • Retinitis Pigmentosa • Cone Rod Dystrophy • Choroideremia 	Intelligent Retinal Implants System (IRIS 2)	Pixium Vision SA	Europe	Not applicable	NCT02670980	Active, not recruiting	April 2022
• Choroideremia	44Ch Bionic Eye Device	Mobius Medical Pty Ltd.	Australia	Not applicable	NCT03406416	Enrolling by invitation	December 2020

Non-interventional natural history studies for people with Inherited Retinal Diseases

Inherited Retinal Disease	Study Sponsor	Country	Study name	ClinicalTrials.gov Identifier	Recruitment status	Estimated primary study completion date
All IRDs	Foundation Fighting Blindness Clinical Research Institute	USA	Inherited Retinal Degenerative Disease Registry	NCT02435940	Recruiting	June 2037
Leber Hereditary Optic Neuropathy	GenSight Biologics	USA	REALITY LHON Registry (REALITY)	NCT03295071	Recruiting	March 2019
<ul style="list-style-type: none"> • Leber Hereditary Optic Neuropathy • Kearns-Sayre syndrome 	Columbia University National Institute of Neurological Disorders and Stroke (NINDS)	USA Canada	North American Mitochondrial Disease Consortium Patient Registry and Biorepository (NAMDC)	NCT01694940	Recruiting	December 2025
Leber Congenital Amaurosis 2 (RPE65)	MeiraGTx UK II Ltd	USA UK	MGT005 Natural History Study	NCT02714816	Recruiting	March 2021
Leber Congenital Amaurosis (CEP290)	Editas Medicine, Inc.	USA Germany Netherlands	Natural History Study of CEP290-Related Retinal Degeneration	NCT03396042	Recruiting	August 2019
Usher syndrome	Jaeb Center for Health Research	USA	Rate of Progression in USH2A Related Retinal Degeneration (RUSH2A)	NCT03146078	Recruiting	May 2021
X-linked Retinitis Pigmentosa (RPGR-ORF15)	Applied Genetic Technologies Corp	USA	Clinical Evaluation of Patients With X-linked Retinitis Pigmentosa (XLRP)	NCT03314207	Recruiting	December 2021

<ul style="list-style-type: none"> • Retinitis Pigmentosa • Stargardt's Disease • Cone-rod dystrophy 	National Eye Institute (NEI)	USA	Rod and Cone Mediated Function in Retinal Disease	NCT02617966	Recruiting	August 2020
Choroideremia	Nightstar Therapeutics	USA Brazil Canada Finland France Germany Netherland UK	Natural History of the Progression of Choroideremia Study (NIGHT)	NCT03359551	Recruiting	November 2019
X-Linked Juvenile Retinoschisis	National Eye Institute (NEI)	USA	Clinical and Genetic Studies of X-Linked Juvenile Retinoschisis	NCT00055029	Recruiting	Not specified