Research Funding provided by Choroideremia Research Foundation

Choroideremia RESEARCH FOUNDATION

CureCHM.org

Funded	Researcher Name	Institution	Project Title
2002	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Choroideremia Research Lab Supplies
2003	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Development of CHM Mouse Model
2004	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Generation of CHM Viral Vector, pt. 1
2005	Kirill Alexandrov, PhD	Max Planck Institute, Germany	Forced Expression of REP2 to the Retina
2005	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Generation of CHM Viral Vector, pt. 2
2006	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Preclinical Gene Therapy Study Year 1
2007	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Preclinical Gene Therapy Study Year 2
2010	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA	Mouse Study Testing for Three Viral Vector Candidates
2011	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA	Alternative In-Vitro Assay to Evaluate Three Viral Vector Candidates
2011	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Pre-Clinical Gene Therapy Study Year 3
2012	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA	Purchase of MP-1 Nidek digital retinal microperimeter equipment
2012	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Ataluren to Treat Nonsense-Mediated Choroideremia; Evaluate 6 Readthrough Compounds on Zebrafish and iPS Derived CHM Cell Lines, Fibroblast, RPE with Dr. Kalatzis; Grant 1
2012	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania	First Generation Gene Therapy in Collaboration with Spark Therapeutics, pt. 1
2013	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Pre-Clinical Gene Therapy Studies pt. 2
2013	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Ataluren to Treat Nonsense-Mediated Choroideremia; Evaluate 6 Readthrough Compounds on Zebrafish and iPS Derived CHM Cell Lines, Fibroblast, RPE with Dr. Kalatzis; Grant 2

Funded	Researcher Name	Institution	Project Title
2013	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA	First Generation Gene Therapy in Collaboration with Spark Therapeutics, pt.2
2013	Vasiliki Kalatzis, PhD, Human Genetics, HDR Life Sciences	Institute for Neurosciences of Montpellier, INSERM, France	Pre-Clinical Gene Therapy Studies for Choroideremia Using a Human Cellular Model: Differentiation of Patient iPS Cells into Retinal Cells, pt. 2
2013	David Gamm, MD, PhD, Director, McPherson Eye Research Institute, Associate Professor, Ophthalmology and Visual Sciences	University of Wisconsin, Madison, WI	Microscope and Laboratory Equipment for Choroideremia Research
2013	Ian MacDonald, BsC, PhD, Professor of Metabolic Physiology, Faculty of Medicine & Health Sciences	University of Nottingham, UK	An Open Label Clinical Trial of Retinal Gene Therapy for Choroideremia
2014	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Ataluren to Treat Nonsense-Mediated Choroideremia; Evaluate 6 Readthrough Compounds on Zebrafish and iPS Derived CHM Cell Lines, Fibroblast, RPE with Dr. Kalatzis; Grant 3
2014	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Ataluren to Treat Nonsense-Mediated Choroideremia; Evaluate 6 Readthrough Compounds on Zebrafish and iPS Derived CHM Cell Lines, Fibroblast, RPE with Dr. Kalatzis; Grant 4
2014	Vasiliki Kalatzis, PhD, Human Genetics, HDR Life Sciences	Institute for Neurosciences of Montpellier, INSERM, France	Pre-Clinical Gene Therapy Studies for Choroideremia Using a Human Cellular Model: Differentiation of Patient iPS Cells into Retinal Cells, pt. 1
2014	David Gamm, MD, PhD, Director, McPherson Eye Research Institute, Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	The Potential Role of hiPSCs in the Treatment of Choroideremia
2015	Jean Bennett, MD, PhD, F.M. Kirby Professor of Ophthalmology	Scheie Eye Institute, Perelman School of Medicine, University of Pennsylvania	Multi-Focal ERG/Visual Evoked Potentials Machine
2015	Mark Pennesi, MD, PhD Assistant Professor in Ophthalmic Genetics	Oregon Health and Science University, Portland, OR	Exploring the Potential of OCT Angiography to Monitor Progression in Choroideremia
2015	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Direct Reprogramming of Fibroblasts into Functional RPE Cells by Specific Transcription Factors
2015	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Whole Organism Screening for Protective/Regenerative Drug Therapeutics in the CHM Zebrafish Model
2015	Jeffrey S. Mumm, PhD, Helen Larson & Charles Glenn Grover Professor in Ophthalmology, Associate Professor of Ophthalmology	Wilmer Eye Institute, Johns Hopkins Medicine, Baltimore, MD	Whole Organism Screening for Protective/Regenerative Drug Therapeutics in the CHM Zebrafish Model
2015	Gerald Lutty, PhD, Director, Ocular Vasculogenesis and Angiogenesis Laboratory; Professor of Ophthalmology	Wilmer Eye Institute, Johns Hopkins Medicine, Baltimore, MD	Production and Testing of CHM hiPSC-Derived Retinal and Vascular Cells (part 1)

Funded	Researcher Name	Institution	Project Title
2015	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	Establishment of CHM Biobank
2015	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	Production and Testing of CHM hiPSC-Derived Retinal and Vascular Cells (part 2)
2015	n/a	4D Molecular Therapeutics, Emeryville, CA	Development of AAV Capsid Variants with Enhanced Pan retinal Gene Delivery of the REP-1 Transgene for the Treatment of Choroideremia
2016	Edwin Stone, MD, PhD, Seamans-Hauser Chair in Molecular Ophthalmology; Director, Molecular Ophthalmology Laboratory; Director, Carver Family Center for Macular Degeneration; Director, Carver Nonprofit Genetic Testing Laboratory; Director, Institute for Vision	University of Iowa Foundation, Iowa City, IA	Project CHM Genotyping Program (part 2) - Funded in Conjunction with PTC Therapeutics
2016	Robert MacLaren, MB, ChB, Dphi, FRCOphth, FRCS, FACS, FMedSci, Professor of Opthamology	University of Oxford, UK	OPI Lumera OCT Microscope Equipment Purchase
2016	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Whole Organism Screening for Protective/Regenerative Drug Therapeutics in the CHM Zebrafish Model; grant 2
2016	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Freezer for CHM Research Samples
2016	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	Investigating the Degenerating Choroid in Choroideremia
2017	Michael Young, PhD, FARVO, Co-Director, Ocular Regenerative Medicine Institute and Director, Minda de Gunzburg Center for Retinal Regeneration	Schepens Eye Institute, Mass General Boston; Department of Opthamology, Harvard Medical School, Boston, MA	Localized Gene Delivery Through Suprachoroidal Space Using a Novel Auto Stop Needle
2018	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	Year 1: Elucidating the Function of REP1 in Human Pluripotent Stem Cell-Derived RPE and Photoreceptor cells – funded in partnership with the Choroideremia Research Foundation Canada
2018	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	Determining the Downstream Consequences of Endogenous REP1 Activity in Human RPE and Photoreceptor cells
2018	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	How CHM Defect Affects Cross Talk Between Organelles and Cellular Functions such as Mitochondria, Lysosome, Autophagy, and Proteostasis

Funded	Researcher Name	Institution	Project Title
2018	Keirnan Willett, MD, Department of Ophthalmology	University of Pennsylvania, Philadelphia, PA	Vascular Biomarkers in Retinal Gene Therapy for Leber Congenital Amaurosis and Choroideremia - funded in partnership with Fight for Sight
2018	Jason A. Mills, PhD, Research Investigator and Kathleen Boesze-Battaglia, PhD, Professor of Biochemistry and Biophysics	MDBR, Orphan Disease Center, University of Pennsylvania, Philadelphia, PA	Targeting Phagosome Maturation to Restore Dysfunctional Retinal Pigmented Epithelium in CHM – funded in partnership with the Penn Orphan Disease Center
2019	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	Waisman Center, University of Wisconsin, Madison, WI	Year 2: Elucidating the Function of REP1 in Human Pluripotent Stem Cell-Derived RPE and Photoreceptor Cells
2019	Miguel Seabra, MD, PhD, Professor, CEDOC, Chronic Diseases Research Center	Nova Medical School, University of Lisbon, Portugal	Mechanisms in Cell Death in Choroideremia
2020	Katrina Stingl, MD, Ophthalmologist, Clinical Scientist	University Eye Hospital, Tübingen, Germany	Adaptive Optics Imaging in Follow-Ups of Choroideremia Patients after Gene Therapy- funded in partnership with the Penn Orphan Disease Center
2020	Richard Harbottle, PhD, Group Leader, DNA Vector Group Leader	German Cancer Research Centre, DKFZ, Heidelberg, Germany	Autonomously Replicating DNA Nanovectors for Gene and Cell Therapy of Choroideremia
2020	David Williams, PhD, Professor in Residence, Ophthalmology	University of California, Los Angeles, CA	Understanding Mitochondrial Defects in Choroideremia
2020	Kim Edwards, Graduate Student	University of Wisconsin, McPherson Eye Research Institute, Madison, WI	RANDY WHEELOCK RESEARCH AWARD WINNER: Identifying the Function of REP-1 Protein in Retina (RPE/Photoreceptors) and Non-Retina Tissues
2020	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	University of Wisconsin, McPherson Eye Research Institute, Madison, WI	Randy Wheelock Research Award Budget Supplement
2020	Abigail Fahim, MD, PhD, Clinical Assistant Professor, Ophthalmology and Visual Sciences	Kellogg Eye Center, University of Michigan, Ann Arbor, MI	Investigating Choroideremia Pathophysiology using iPSC-derived Retinal Pigment Epithelium – funded in partnership with the Choroideremia Research Foundation Canada
2020	Stacey Hume, PhD, FCCMG, Associate Professor, Department of Medical Genetics	University of Alberta, Canada	BOREN FAMILY RESEARCH AWARD: Identifying the Cause of a Discordant Phenotype in Two Brothers with the Identical CHM Mutation – funded in partnership with the Choroideremia Research Foundation Canada
2020	Yi (Fay) Zhai, MD, PhD, Clinical Research Fellow, Department of Ophthalmology	University of Alberta, Canada	OSTER FAMILY RESEARCH AWARD: Measuring the En Face Ellipsoid Zone (EZ) Area as a Biomarker of Photoreceptor Structure/Function in Choroideremia – funded in partnership with the Choroideremia Research Foundation Canada

Funded	Researcher Name	Institution	Project Title
2021	Mariya Moosajee, MBBS, BsC (Hons), PhD, FRCOphth, Consultant Ophthalmic Surgeon and Clinical Academic Ophthalmologist	University College, London, UK	SALOIS FAMILY RESEARCH AWARD: Neuroprotection for Choroideremia - – funded in partnership with the Choroideremia Research Foundation Canada
2021	Vasiliki Kalatzis, PhD, Human Genetics, HDR Life Sciences	Institute for Neurosciences of Montpellier, INSERM, France	GLEASON FAMILY RESEARCH AWARD: A Novel Approach to Unravelling the Pathophysiology of CHM using iPSC-derived RPE from Patients- funded in partnership with the Choroideremia Research Foundation Canada
2021	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	University of Wisconsin, McPherson Eye Research Institute, Madison, WI	THE AUBURN THETA CHI, CHI CHAPTER RESEARCH AWARD: Generation of Human iPSC Lines with Patient-Relevant REP-1 Mutation
2021	Ivan Conte, PhD, Assistant Professor, Department of Biology, Polytechnic and Basic Sciences School	University of Naples Federico II, Italy	Pharmacological induction of autophagy to treat CHM – funded in partnership with the Penn Orphan Disease Center
2021	Jasleen Kaur Jolly MSc BSc (Hons) MCOptom, Senior Clinical Research Fellow	University of Oxford, Nuffield Department of Clinical Neurosciences, UK	RICKETTS FAMILY RESEARCH AWARD: The Visual Brain in Choroideremia
2021	Cynthia Qian, MD, FRCSC, DABO, Clinical Assistant Professor	University of Montreal, Canada	RANDY WHEELOCK RESEARCH AWARD WINNER: Characterizing the phenotypical findings in female carriers with confirmed CHM mutation using multimodal imaging and functional testing; funded in partnership with the Choroideremia Research Foundation Canada
2021	Abigail Fahim, MD, PhD Clinical Assistant Professor, Ophthalmology and Visual Sciences	Kellogg Eye Center, University of Michigan, Ann Arbor, MI	Investigating Choroideremia Pathophysiology using iPSC-derived Retinal Pigment Epithelium – year 2 – funded in partnership with the Choroideremia Research Foundation Canada
2021	Bhanu P. Telugu, DVM, PhD, President & CSO	RenOVAte Biosciences, Inc.	CHM Porcine Animal Model Development – funded in partnership with Choroideremia Research Foundation Canada
2022	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	University of Wisconsin, McPherson Eye Research Institute, Madison, WI	Assessing the potential of engineered tRNA readthrough technology to restore Rab Escort Protein-1 (REP-1) protein expression-- funded in partnership with the Penn Orphan Disease Center
2022	David Gamm, MD, PhD, Director, McPherson Eye Research Institute; Associate Professor, Ophthalmology and Visual Sciences	University of Wisconsin, McPherson Eye Research Institute, Madison, WI	EINHORN FAMILY RESEARCH AWARD: MDBR Supplement: Assessing the potential of engineered tRNA readthrough technology to restore Rab Escort Protein-1 (REP-1) protein expression
2022	Vasiliki Kalatzis, PhD, Human Genetics, HDR Life Sciences	Institute for Neurosciences of Montpellier, INSERM, France	Unravelling the Pathophysiology of CHM using innovative approaches – funded in partnership with Choroideremia Research Foundation Canada
2022	Ian MacDonald, MSc, MD, CM, Professor Emeritus, Department of Ophthalmology and Visual Sciences	University of Alberta, Canada	Developing an antisense oligonucleotide therapy for choroideremia – funded in partnership with the Choroideremia Research Foundation Canada

Funded	Researcher Name	Institution	Project Title
2022	Sena Gocuk, B.Sci (Hons), D.Optom, M.Phil, Department of Optometry and Vision Sciences	University of Melbourne, Australia	Structural and functional changes in female carriers of choroideremia: A longitudinal study
2022	Jasleen Jolly, DPhil, MSc, BSc (Hons), MCOptom, Associate Professor, Vision and Eye Research Institute	Anglia Ruskin University, Cambridge, UK	MAIA Scotopic Microperimetry Equipment Purchase
2022	Bhanu P. Telugu, DVM, PhD, President & CSO	RenOVAte Biosciences, Inc.	RANDY WHEELOCK RESEARCH AWARD WINNER: CHM Porcine Animal Model Development year 2