

Maria Carolina Ortube, MD

Curriculum Vitae

2008-2015:

Director of Clinical Research Studies
Retinal Disorders & Ophthalmic Genetics Division
Jules Stein Eye Institute, UCLA
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2015: Instituto de la Vision
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Education

1990-1997: Medical School: Buenos Aires University, Hospital de Clinicas José de San Martín. Buenos Aires, Argentina.
Degree: Medical doctor.

1998-2001: Ophthalmology Residency Program: Centro de Ojos Quilmes. Buenos Aires, Argentina.
Degree: Ophthalmologist

1999–2001: Ophthalmology Postgraduate Course: Hospital de Clinicas José de San Martín.
Degree: Ophthalmologist. National certification by Buenos Aires University. Argentina.

2001-2002: Juan Garrahan Hospital. Pediatric Ophthalmology Department. Buenos Aires, Argentina.

2002-2005: Pediatric Ophthalmology and Strabismus Research Fellowship Visiting Assistant Professor. Jules Stein Eye Institute, UCLA. Los Angeles, California.
Joseph L. Demer, MD, PhD; Sherwin Isenberg, MD; Arthur Rosenbaum, MD.

Licensure

1998- Present: National Ministry of Health, MN 98.362, for practice of Medicine. Argentina.

2001- Present: Ophthalmologist.

2007: Spanish Ministry of Health, for practice of Medicine. Madrid, Spain.

2009- Present: National certification. Buenos Aires University. Argentina.
(*Especialista Universitario en Oftalmología*)

Citizenship

Argentinian citizen

USA permanent resident

Research and/or professional experience

1998-2001: Resident physician in Ophthalmology. Centro de Ojos Quilmes, Buenos Aires, Argentina.

2001-2002: Fellow. Juan Garrahan National Pediatric Hospital, Buenos Aires, Argentina.

2002-2005: Research Fellow in Pediatric Ophthalmology & Strabismus, Jules Stein Eye Institute & Harbor UCLA Medical Center.

2005-2006: Postdoctoral Fellow in Ocular Albinism. Basic science.

2006-2008: Clinical research. Jules Stein Eye Institute, UCLA.

2008- 2015: Director of Clinical Research Studies. Retinal Disorders & Ophthalmic Genetics Division.

2015: Instituto de la Vision. Buenos Aires. Argentina.

Societies

Association of Research in Vision and Ophthalmology. Member.

Pan American Medical Association. Member

World Society of Pediatric Ophthalmology & Strabismus. Member

CLADE (Consejo Latinoamericano de Estrabismo) Member

AMD Gene Consortium

Honors & Awards

2003: Clinical Fellow Award: Pediatric Ophthalmology and Strabismus Research. Jules Stein Eye Institute, University of California.

September 2011: O-1 Visa for Individuals with extraordinary abilities in sciences. (Granted by United States Citizenship & Immigration Services, USCIS).

May 2014: Milstein Award. Ministry of Science, Technology and Productive Innovation. Argentina.

June 2015: Troutman-Véronneau Award
Pan American Association of Ophthalmology
Fusion after treatment of strabismus secondary to brain lesions.

Grants

2004: Knights Templar Foundation Research Grant.
Principal investigator. Project: Orbital imaging in Craniosynostosis, understanding the pathophysiology of complex strabismus.

2005: Vision of Children
Principal Investigator. Project: Ocular Albinism.

2010-2013: NIH SBIR 2R44 EY-018025-02
Co-investigator: Developing a noninvasive method and device for assessing the degree of midperipheral retinal ischemia in diabetic retinopathy.

Editorial Services to Scholarly Publications

2010- Present: Reviewer for *British Journal of Ophthalmology*.

2010- Present: Reviewer for *Current Eye Research*.

2010- Present: Reviewer for *Experimental Eye Research*.

2011- Present: Reviewer for *Ophthalmology*.

2012- Present: Reviewer for *American Journal of Medical Genetics*.

2014- Present: Reviewer for *Journal of the American Association for Pediatric Ophthalmology & Strabismus*.

2014- Present: Reviewer for *Cancer Informatics*.

2014: Present: Editorial Board Member: *Revista de Genetica Medica*. Spain.

Teaching

2007-2012: Assisted residents in Grand rounds presentations at Jules Stein Eye Institute.

2007: Co-Mentored a medical student from New York University: Matej Polomsky. (Publication #13)

2009-2011: Senior Instructor: Ocular Genetics Conferences. Jules Stein Eye Institute.

2010: Co-Mentored a medical student from UCLA: Daniel Choi. (Publication # 8)

2012-2013: Mentored Victor Chen, undergraduate student and Van Lam, clinical coordinator. Both candidates were accepted to present at the 2013 ARVO conference in Seattle, WA. (Presentations 33 & 34).

2014: Lecturer. Leber Congenital Amaurosis: Genetics. Congreso Neurooftalmologica XXII. October 11th 2014. Buenos Aires. Argentina.

2014: Curso de Postgrado en Genética Médica. Lecturer. University of Valencia, Spain. Ophthalmic Genetics. November 6th 2014. Valencia. Spain.

url: <http://medicinagenomica.com/curso-genetica-medica/online/#profesorado>

Protocols Approved for Human Investigation at UCLA

- 1) Fusion after treatment of patients with acquired strabismus due to brain lesions. (Principal Investigator).
- 2) Biomechanical analysis in strabismus surgery. (Co-investigator).
- 3) Understanding the genetics of inherited eye disorders. (Co-investigator).
- 4) Hereditary ocular conditions: retrospective study to correlate phenotype-genotype association. (Co-investigator).
- 5) Clinical characterization, genetic testing and visual function in patients with Stargardt disease. (Co-investigator).
- 6) Rapid, non-invasive, regional functional imaging of the retina. (Co-

investigator).

7) GARM: Genetics of age related maculopathy. (Co-investigator)

Meeting Presentations and Lectures

1) Poster: Corneal nerve development after refractive surgery and penetrating keratoplasty. XXV Annual Course. Argentine Foundation of Ophthalmology. October 14-16 1998. Maria Carolina Ortube, MD, Rendo Javier, MD, Valvecchia Gerardo, MD- Albertazzi Roberto, MD. Centro de Ojos Quilmes.

2) Scientific paper: Corneal nerve development after refractive surgery and penetrating keratoplasty. Maria Carolina Ortube, MD, Rendo Javier, MD, Valvecchia Gerardo MD. March 1999. Refractive Surgery magazine review.

3) Poster: Heredity of Rieger Syndrome. Argentine Congress of Ophthalmology. June 6-10 1999. Maria Carolina Ortube, MD, Rendo Javier, MD, Iurescia Alejandra, MD, Izaguirre Amelia, MD. Centro de Ojos Quilmes.

4) Poster: Ocular toxicity of chloroquine. XXVI Annual Course, Argentine Foundation of Ophthalmology, 1999. Maria Carolina Ortube, MD, Ciliberti Susana MD, Sapia Veronica, MD, Langman Elena, MD. Centro de Ojos Quilmes, Ramos Mejia Hospital, Fernandez Hospital. Buenos Aires, Argentina.

5) Poster: Amaurosis by methanol. Congress of the Argentine Society of Ophthalmology. Maria Carolina Ortube, MD, Zurovesky Estela, MD, Zanutigh Virginia, MD. July 2000. Hospital Evita Pueblo. Buenos Aires, Argentina.

6) Symposium: Keratoconus. Argentine Society of Refractive Surgery. Albertazzi Roberto, MD and Ortube Maria Carolina, MD. Buenos Aires, Argentina. November 18th 2000.

7) Lecture: Prospective, Clinical and epidemiological study on thirty families with keratoconus: Pan American Congress of Ophthalmology. July 22-25 2001. Buenos Aires, Argentina.

8) Lecture: Keratoconus. Clinical and topographic characteristics in thirty families in Argentina: July 22-25 2001. Maria Carolina Ortube, MD, Albertazzi Roberto MD. Pan American Congress of Ophthalmology. Buenos Aires, Argentina.

9) Poster: Unusual presentation of keratoconus in children. Maria Carolina Ortube, MD, Londero Lisa, MD. Centro de Ojos Quilmes. Pan American Congress of Ophthalmology. July 22-25 2001. Buenos Aires, Argentina.

10) Poster: Neurofibromatosis. Familiar Case. Rendo Javier, MD; Maria Carolina Ortube, MD. Centro de Ojos Quilmes. Pan American Congress of Ophthalmology. July–25 2001. Buenos Aires, Argentina.

11) Poster: Pseudotumor cerebri induced by pregnancy. Estela Zurovesky, MD, Maria Carolina Ortube, MD, Miriam Perina, MD; Carlos Magnoni, MD; Maria Olgado, MD. Pan American Congress of Ophthalmology. July 22-25 2001. Buenos Aires, Argentina.

12) Lecture: Orbital Imaging Demonstrates Occult Blowout Fractures in Complex Strabismus Cases. Maria Carolina Ortube, MD; Joseph Demer, MD; Arthur Rosenbaum, MD. March 2003. AAPOS Meeting. (American Academy of Pediatric Ophthalmology and Strabismus). Hawaii.

13) Lecture: Orbital Imaging demonstrates Occult Blowout fractures in Complex Strabismus. Maria Carolina Ortube MD, Arthur L Rosenbaum MD, Joseph Demer MD, PhD. May 31, 2003. Post ARVO meeting: Jules Stein Eye Institute.

14) Poster: Orbital Magnetic Resonance Imaging of Extraocular Muscles in Chronic Progressive External Ophthalmoplegia: Specific Diagnostic Findings. Maria Carolina Ortube, MD, Rahul Bhola, MD, Joseph L. Demer, MD, PhD. March 27-31, 2004. AAPOS Meeting. Washington DC.

15) Lecture: Orbital Magnetic Resonance Imaging (MRI) of Extraocular Muscles (EOMs) Elucidates Mechanism of Ptosis and Strabismus in Jacobsen Syndrome. Maria Carolina Ortube, Joseph L. Demer. April 25- 29, 2004. Association for Research in Vision and Ophthalmology (ARVO) Meeting. Fort Lauderdale, Florida.

Invest Ophthalmol Vis Sci 2004; 45: E-Abstract 4577.

16) Poster: Magnetic Resonance Imaging (MRI) Demonstrates the Endophenotype of Duane Retraction Syndrome (DRS) Linked to the DURS2 Locus. J.L. Demer, M.C. Ortube, S. Karim, C. Andrews and E.C. Engle. Jules Stein Eye Institute, Los Angeles, CA. Boston Children's Hospital, Boston, MA. April 25-29, 2004. Association for Research in Vision and Ophthalmology (ARVO) Meeting. Fort Lauderdale, Florida.

Invest Ophthalmol Vis Sci 2004; 45: E-Abstract 4574.

17) Poster: Orbital Magnetic Resonance Imaging of Extraocular Muscles in Chronic Progressive External Ophthalmoplegia: Specific Diagnostic Findings. May 21-22, 2004. Post ARVO meeting: Program Participant. Jules Stein Eye Institute.

18) Poster: Fusion after treatment of patients with acquired strabismus due to brain lesions. Maria Carolina Ortube, MD, Federico G. Velez, MD, Joseph L.

Demer, MD, PhD, Arthur L. Rosenbaum, MD. March 9-13, 2005: AAPOS Meeting. Orlando, Florida.

19) Lecture: Strabismus patterns in craniofacial anomalies. JSEI Clinical and Research Seminar. May 20, 2005. Los Angeles, California.

20) Poster: Two separate mutations contributing to disease severity in an autosomal dominant Retinitis Pigmentosa (RP) family. Michael B. Gorin, MD, PhD, A. Martinez, CGC, Maria Carolina Ortube, MD, E. Spector, PhD, P. Chiang, PhD. American Society of Human Genetics, November 2008.

21) Lecture: Ocular genetics conference at Jules Stein Eye Institute: Osteoporosis pseudoglioma syndrome. January 2009.

22) Poster: Further explorations in the genetics of Age-related Maculopathy (ARM). Michael B. Gorin, MD, PhD, Johanna Jakobsdottir, PhD, Yvette Conley, PhD, M. Carolina Ortube, MD, Robert E. Ferrell, PhD, Wei Cheng, PhD, D. Phil, Anand Swaroop, PhD, Albert O. Edwards, MD, PhD, Dwight E. Stambolian, MD, PhD, Daniel Weeks, PhD. ARVO meeting (Association for Research in Vision and Ophthalmology), May 2009.

23) Lecture: Ocular genetics conference: Vascular angiomas. May 2009.

24) Lecture: Comparative regional pupillography as a noninvasive screening method for diabetic retinopathy. Maria Carolina Ortube, MD, Fei Yu, PhD, Alex Kiderman, PhD, Michael B. Gorin, MD, PhD. Jules Stein Clinical Research Seminar, June 12th 2009.

25) Lecture: Ocular genetics conference at Jules Stein Eye Institute: Fraser syndrome. October 2009.

26) Lecture: Ocular genetics conference at Jules Stein Eye Institute: Genetics of congenital fibrosis of extraocular muscles (CFEOM). December 2009

27) Poster: GARM II Study: A Web-based Study on the Genetics of Age-related Macular Degeneration. Maria Carolina Ortube, MD, Yvette Conley, PhD; Daniel Weeks, PhD; Ariadna Martinez, CGC, Heather Leatherwood, BS; Arturo Garcia, BS; Steve Schmidhauser; Carlos Carballo, BS and Michael B. Gorin, MD, PhD. Jules Stein Eye Institute Vision Science conference. October 2010.

28) Poster: Genetic ME - A visualization application for merging and editing pedigrees for genetic studies. D. K. Bui, M. C. Ortube, MD, D. E. Weeks, PhD, A. Martinez, MS, MS, Y. Conley, PhD, M. B. Gorin, MD, PhD. American Society of Human Genetics. November 2010

29) Poster: Molecular characterization of a cohort of Stargardt and Stargardt-like

affected individuals and families. M. B. Gorin, A. Martinez, YQ. Gao, M. C. Ortube, D. B. Farber. American Society of Human Genetics. November 2010

30) Lecture: Strabismus due to brain lesions and central fusion disruption: Long-term follow-up in a cohort of 50 patients. AAPOS (American Academy of Pediatric Ophthalmology & Strabismus). San Diego, March 2011

31) Poster: Orbital Imaging In Strabismus Secondary To Saethre Chotzen Syndrome. ARVO 2011 Visionary Genomics (Association for Research in Vision and Ophthalmology). Fort Lauderdale, May 2011. M. Carolina Ortube, Joseph L. Demer. Jules Stein Eye Institute, UCLA.

Invest Ophthalmol Vis Sci 2011; 52: E-Abstract 6377.

32) Lecture: Homozygosity for ABCA4 R602W missense mutation in a pediatric patient with rapidly progressive retinal dystrophy. Medical Retina Symposium: Second World Conference of Pediatric Ophthalmology & Strabismus. Lecturer. Milan, Italy. September 2012.

33) Poster: The Genetics of Age-Related Maculopathy II (GARM II) study: an interim analysis. Lam, Van; Ortube, Maria Carolina; Conley, Yvette; Weeks, Daniel E.; Martinez, Ariadna; Aguilar, Nelson J.; Andon, Victor; Machuca, Orlando; Su, Angela; Gorin, Michael B. ARVO (Association for Research in Vision and Ophthalmology). Seattle, WA. May 5th 2013.

Invest Ophthalmol Vis Sci 2013; 54: E-Abstract 231.

34) Poster: Diagnostic disparities in testing for hydroxychloroquine (HCQ)-related ocular toxicity. Chen, Victor A.; Ortube, Maria Carolina; Nusinowitz, Steven; Gorin, Michael B. ARVO (Association for Research in Vision and Ophthalmology). Seattle, WA. May 6th 2013.

Invest Ophthalmol Vis Sci 2013; 54: E-Abstract 1564.

35) Poster: Orbital imaging in craniosynostosis: Understanding the pathophysiology of complex strabismus. Ortube, Maria Carolina; Velez, Federico G.; Demer, Joseph L. ARVO (Association for Research in Vision and Ophthalmology). Seattle, WA. May 6th 2013.

Invest Ophthalmol Vis Sci 2013; 54: E-Abstract 1936.

36) Poster: Homozygosity for ABCA4 R602W missense mutation in a pediatric patient with rapidly progressive retinal dystrophy. Jules Stein Eye Institute, Vision Science Conference. Lake Arrowhead, California. October 27th, 2013.

37) Poster. ARVO (Association for Research in Vision and Ophthalmology). Title: " The Challenges, limitations and uncertainties of "whole" exome sequencing (WES) for hereditary ocular disorders". Ortube, Maria Carolina; Strom, Sam; Martinez Ariadna; Gorin Michael B. Orlando, Florida. May 8th 2014.

38) Lecture: Title: " Senior Loken syndrome: Leber congenital amaurosis and nephronophthisis ". Maria Carolina Ortube, MD; Michael B. Gorin, MD, PhD. Argentine Congress of Ophthalmology. Buenos Aires, Argentina. May 14-17, 2014.

39) Lecture: Title: " The Challenges, limitations and uncertainties of "whole" exome sequencing (WES) for hereditary ocular disorders". Maria Carolina Ortube, MD; Sam Strom, PhD; Ariadna Martinez, CGC; Michael B. Gorin, MD, PhD. Argentine Congress of Ophthalmology. Buenos Aires, Argentina. May 14-17, 2014.

40) 2014: Lecture. Leber Congenital Amaurosis: Genetics. Congreso Neurooftalmologica XXII. October 11th 2014. Buenos Aires. Argentina.

41) Lecture: Title: Ophthalmic Genetics. Curso de Postgrado en Genética Médica. Lecturer. (Profesorado). November 6, 2014. University of Valencia, Spain.

42) Genetics-based testing of functional and structural endophenotypes for pre- and early-age-related macular degeneration (AMD). Conference: Arnold and Mabel Beckman Initiative for Macular Research. Michael B. Gorin, MD, PhD; Maria Carolina Ortube, MD; Steven Nusinowitz, PhD. January 15-17 2015. Beckman Center of the National Academies of Sciences and Engineering, Irvine, California.

43) Lecture: Update on genetics of AMD and biomarkers. March 19, 2015. Argentine Congress of Ophthalmology. Buenos Aires. Argentina.

44) Lecture: Stargardt disease. April 10, 2015. RETINAR. Argentine Society of Retina and Vitreous. National Academy of Medicine. Buenos Aires, Argentina.

45) Lecture: Clinical trials oriented to the treatment of heredodegenerative conditions. April 10, 2015. RETINAR. Argentine Society of Retina and Vitreous. National Academy of Medicine. Buenos Aires, Argentina.

46) Lecture: Genetic tests in Argentina. May 14, 2015. Argentine Council of Ophthalmology. Buenos Aires, Argentina.

47) Future Lecture: Fusion after treatment of patients with acquired strabismus due to brain lesions. 2015 Troutman-Véronneau Prize. August 5, 2015. Pan American Association Congress. Bogotá. Colombia.

48) Future Lecture: Genetics in Ophthalmology. August 21, 2015. RETINAR. Argentine Society of Retina and Vitreous. National Academy of Medicine.

Buenos Aires, Argentina.

49) Future Lecture: Three atypical phenotype-genotype associations in paediatric cases. September 4, 2015. World Congress of Paediatric Ophthalmology & Strabismus. Barcelona, Spain.

50) Futura Lecture: Ocular manifestations in craniofacial patients. Sohag University. Egypt. October 2015.

51) Future Lecture: The ophthalmologist's role in the craniofacial team. Sohag University. Egypt. October 2015.

52) Future Lecture: Genetics in Ophthalmology. Postgraduate Course in Medical Genetics. Valencia University, Spain . October or November 2015. Valencia, Spain.

Publications

1) Ortube MC, Rosenbaum AL, Goldberg RA, Demer JL. Orbital imaging demonstrates occult blowout fracture in complex strabismus. J. AAPOS. 2004 Jun; 8 (3) 264-73.

2) Bholra R, Rosenbaum AL, Ortube MC, Demer JL. High-resolution magnetic resonance imaging demonstrates varied anatomic abnormalities in Brown syndrome. J. AAPOS. 2005 Oct; 9 (5) 438-48.

3) Demer JL, Ortube MC, Engle EC, Thacker N. High-resolution magnetic resonance imaging demonstrates abnormalities of motor nerves and extraocular muscles in patients with neuropathic strabismus. J.AAPOS 2006 Apr; 10 (2) 135-42.

4) Ortube MC, Bholra R, Demer JL. Orbital magnetic resonance imaging of extraocular muscles in chronic progressive external ophthalmoplegia: specific diagnostic findings. J.AAPOS 2006 Oct; 10 (5) 414-8.

5) Kau HC, Tsai CC, Ortube MC, Demer JL. High-resolution magnetic resonance imaging of the extraocular muscles and nerves demonstrates various etiologies of third nerve palsy. Am. J. Ophthalmol. 2007 Feb; 143(2) 280-287.

6) Ortube MC, Lazareff J, Vinters HV, Velez FG. Orbital ectopic brain tissue in Aicardi syndrome. J. Craniofac. Surg. 2010 Sep; 21(5): 1551-3.

7) Ortube MC, Dipple K, Setoguchi Y, Kawamoto HK Jr, Demer JL. Ocular manifestations of oblique facial clefts. J. Craniofac. Surg. 2010 Sep; 21(5): 1630-1.

8) Choi D, Ortube MC, McCannel CA, Sarraf D, Hubschman JP, McCannel TA, Gorin MB. Sustained Elevated Intraocular Pressures after Intravitreal Injection of Bevacizumab, Ranibizumab, and Pegaptanib. *Retina*. 31(6): 1028-1035, June 2011.

9) Sofat R, Casas JP, Webster AR, Bird AC, Mann SS, Yates JR, Moore AT, Sepp T, Cipriani V, Bunce C, Khan JC, Shahid H, Swaroop A, Abecasis G, Branham KE, Zarepari S, Bergen AA, Klaver CC, Baas DC, Zhang K, Chen Y, Gibbs D, Weber BH, Keilhauer CN, Fritsche LG, Lotery A, Cree AJ, Griffiths HL, Bhattacharya SS, Chen LL, Jenkins SA, Peto T, Lathrop M, Leveillard T, Gorin MB, Weeks DE, Ortube MC, Ferrell RE, Jakobsdottir J, Conley YP, Rahu M, Seland JH, Soubrane G, Topouzis F, Vioque J, Tomazzoli L, Young I, Whittaker J, Chakravarthy U, de Jong PT, Smeeth L, Fletcher A, Hingorani AD. Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. *Int J Epidemiol*. 41 (1): 250-62. 2012

10) Strom SP, Gao YQ, Martinez A, Ortube C, Chen Z, Nelson SF, Nusinowitz S, Farber DB, Gorin MB. Molecular diagnosis of putative Stargardt Disease probands by exome sequencing. *BMC Med Genet*. 2012 Aug 3; 13: 67.

11) Ortube MC, Kiderman A, Eydelman Y, Yu F, Aguilar N, Nusinowitz S, Gorin MB. Comparative regional pupillography as a noninvasive biosensor screening method for diabetic retinopathy. *Invest Ophthalmol Vis Sci*. 2013 Jan 2; 54(1): 9-18. doi: 10.1167/iov.12-10241.

12) Fritsche LG, Chen W, Schu M, Yaspan BL, Yu Y, Thorleifsson G, Zack DJ, Arakawa S, Cipriani V, Ripke S, Igo RP Jr, Buitendijk GH, Sim X, Weeks DE, Guymer RH, Merriam JE, Francis PJ, Hannum G, Agarwal A, Armbrecht AM, Audo I, Aung T, Barile GR, Benchaboune M, Bird AC, Bishop PN, Branham KE, Brooks M, Brucker AJ, Cade WH, Cain MS, Campochiaro PA, Chan CC, Cheng CY, Chew EY, Chin KA, Chowers I, Clayton DG, Cojocar R, Conley YP, Cornes BK, Daly MJ, Dhillon B, Edwards AO, Evangelou E, Fagerness J, Ferreyra HA, Friedman JS, Geirsdottir A, George RJ, Gieger C, Gupta N, Hagstrom SA, Harding SP, Haritoglou C, Heckenlively JR, Holz FG, Hughes G, Ioannidis JP, Ishibashi T, Joseph P, Jun G, Kamatani Y, Katsanis N, Keilhauer C, Khan JC, Kim IK, Kiyohara Y, Klein BE, Klein R, Kovach JL, Kozak I, Lee CJ, Lee KE, Lichtner P, Lotery AJ, Meitinger T, Mitchell P, Mohand-Saïd S, Moore AT, Morgan DJ, Morrison MA, Myers CE, Naj AC, Nakamura Y, Okada Y, Orlin A, Ortube MC, Othman MI, Pappas C, Park KH, Pauer GJ, Peachey NS, Poch O, Priya RR, Reynolds R, Richardson AJ, Ripp R, Rudolph G, Ryu E, Sahel JA, Schaumberg DA, Scholl HP, Schwartz SG, Scott WK, Shahid H, Sigurdsson H, Silvestri G, Sivakumaran TA, Smith RT, Sobrin L, Souied EH, Stambolian DE, Stefansson H, Sturgill-Short GM, Takahashi A, Tosakulwong N, Truitt BJ, Tsironi EE, Uitterlinden AG, van Duijn CM, Vijaya L, Vingerling JR, Vithana EN, Webster

AR, Wichmann HE, Winkler TW, Wong TY, Wright AF, Zelenika D, Zhang M, Zhao L, Zhang K, Klein ML, Hageman GS, Lathrop GM, Stefansson K, Allikmets R, Baird PN, Gorin MB, Wang JJ, Klaver CC, Seddon JM, Pericak-Vance MA, Iyengar SK, Yates JR, Swaroop A, Weber BH, Kubo M, Deangelis MM, Léveillard T, Thorsteinsdottir U, Haines JL, Farrer LA, Heid IM, Abecasis GR; AMD Gene Consortium. Seven new loci associated with age-related macular degeneration. *Nat Genet.* 2013 Apr; 45(4): 433-9, 439e1-2. doi: 10.1038/ng.2578. Epub 2013 Mar 3.

13) Gorin MB, Ortube MC, Machuca O, Carballo C, Rupchian A, Wei X, Aguilar N, Polomsky M, Santangelo R, Alvarado I. A novel web-based clinical research tool for a nationwide research study. *J. Clin. Res. Bioethics* 2013 4:145. (Open access)

14) Ortube MC, Strom S, Nelson SF, Nusinowitz S, Martinez A, Gorin MB. Whole exome sequencing detects homozygosity for ABCA4 p.Arg602Trp missense mutation in a pediatric patient with rapidly progressive retinal dystrophy. *BMC Med Genet.* 2014 Jan 20;15(1):11. doi: 10.1186/1471-2350-15-11.

15) Michael B. Gorin, Daniel E. Weeks, Robert V. Baron, Yvette P. Conley, Maria C Ortube, Steven Nusinowitz. Endophenotypes for Age-related Macular Degeneration: Extending our reach into the preclinical stages of disease. *J Clin Med.* 2014 Nov 28;3(4):1335-1356.

16) Bui DK, Jiang Y, Wei X, Ortube MC, Weeks DE, Conley YP, Gorin MB. Genetic ME-a visualization application for merging and editing pedigrees for genetic studies. *BMC Res Notes.* 2015 Jun 16;8:241.