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CURRICULUM VITAE

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EDUCATION AND TRAINING

Undergraduate & Graduate

1987-1993	Istanbul University Istanbul Faculty of Medicine Istanbul, Turkey	MD, 1993	Medicine
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Post-Graduate

1993-1997	Istanbul University Istanbul Faculty of Medicine Department of Ophthalmology Istanbul, Turkey	Residency	Ophthalmology
1999-2002	University of Pittsburgh School of Medicine Department of Ophthalmology Pittsburgh, PA	Fellowship	Ophthalmic Genetics
1999-2002	University of Pittsburgh Graduate School of Public Health Department of Human Genetics Pittsburgh, PA	Fellowship	Clinical Molecular Genetics

APPOINTMENTS AND POSITIONS

Academic

1991	Visiting Researcher	Medical Genetics Division, University of Lausanne, Switzerland
2002-2005	Research Associate	Department of Ophthalmology, UPMC, University of Pittsburgh, Pittsburgh, PA
2005-2006	Research Associate	Department of Human Genetics, GSPH, University of Pittsburgh, Pittsburgh, PA
2006-Present	Research Assistant Professor	Department of Human Genetics, GSPH, University of Pittsburgh, Pittsburgh, PA

Non-Academic

1997-1998	Ophthalmologist	ERMED Eye Center Istanbul, Turkey
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CERTIFICATION AND LICENSURE

Specialty Certification

1997	Ophthalmology	Turkish Board of Ophthalmology
2002	Clinical Molecular Genetics	American Board of Medical Genetics

MEMBERSHIP IN PROFESSIONAL AND SCIENTIFIC SOCIETIES

1994-1998	Turkish Ophthalmological Society (TOD)
2001-2005	Association for Research in Vision and Ophthalmology (ARVO)
2004-2005	American Association for the Advancement of Science (AAAS)
2004-2005	International Society for Genetic Eye Diseases (ISGED)
2000-Present	American Society of Human Genetics (ASHG)

AWARDS AND HONORS

1987	First position by merit in High School of Science graduation
1987	TUBITAK (The Scientific and Technical Research Council of Turkey) Prize, honoring the success rank (2 nd in the region) in National Chemistry Competition
1987-1993	Vehbi Koc Award and Medical School Scholarship, honoring the success rank (21 st among >600,000 participants) in National University Entrance Examination
1993	Second position by merit (out of ~400 graduates) in Medical School graduation, Istanbul Faculty of Medicine
2000-2001	Research Fellowship Grant Award, Fight for Sight, The Research Division of Prevent Blindness America

PROFESSIONAL ACTIVITIES

1. TEACHING

a. Courses Taught

Years Taught	Course Number: Title	Hrs of Lecture, Credits, Enrollment	Role in course
2008	HUGEN 2034: Biochemical and Molecular Genetics of Complex Diseases	3hr/wk, 3 credits, 20-30 students	Lecturer
2009	HUGEN 2034: Biochemical and Molecular Genetics of Complex Diseases	3hr/wk, 3 credits, 20-30 students	Co-Instructor

b. Other Teaching (lectures, tutorials and continuing education courses)

Years Taught	Type of Teaching	Title
2003-2004	Lecturer for the Genetic Counseling Program, GSPH, University of Pittsburgh	‘Ophthalmic Genetics’
2008	Workshop Facilitator, Human Genetics Course, School of Medicine, University of Pittsburgh	‘Inheritance’

c. Co-Advisor for Graduate Student Essays, Theses, and Dissertations

Years Served	Name of Student	Title of Dissertations/Essays/Theses
<i>Ph.D. students:</i>		
2006-2008	Sangita Suresh	“Functional characterization of <i>APOH</i> polymorphisms and their relation to SLE”
2006-2008	Sudeshna Dasgupta	“Association of <i>PON2</i> genetic variation with serum paraoxonase activity and SLE”
2008-Present	Jatinder Singh	“Analysis of <i>ABCA1</i> common and rare variants for their effects on plasma HDL-C levels”
<i>M.S. students:</i>		
2001-2003	Amy Radak	“Mutation screening of <i>RP2</i> and <i>RP3</i> in male patients with XL or isolated forms of RP”
2006-2007	Leia Corthell	“Association of single nucleotide polymorphisms in <i>APOH</i> with SLE”
2006-2008	Sally Hollister	“Sequence variation in <i>APOA2</i> and its relationship with plasma HDL-C levels”
2007-2009	Sarah Hill	“Sequence variation in <i>APOA1</i> and <i>APOA4</i> and their relationship with plasma HDL-C levels”
2008-Present	Kate Sleutz	“Comprehensive evaluation of <i>CD36</i> variants as genetic determinants of plasma HDL-C levels”
2009-Present	Zaheda Radwan	“Effects of <i>APOC4</i> common and rare variants on plasma HDL-C levels”
2009-Present	Dilek Pirim	“Comprehensive analysis of <i>LPL</i> genetic variation in relation to plasma HDL-C levels”

d. Service on Masters or Doctoral Committees

Year Served	Name of Student	Degree Awarded	Title of Dissertations/Essays/Theses
2007	Leia Corthell	M.S.	“Association of SNPs in <i>APOH</i> with SLE”
2008	Sally Hollister	M.S.	“Sequence variation in <i>APOA2</i> and its relationship with plasma HDL-C levels”
2008	Jessica Figgins	M.S.	“Association studies of 22 candidate SNPs with late-onset Alzheimer’s disease”
2008	Sudeshna Dasgupta	Ph.D.	“Association of <i>PON2</i> genetic variation with serum paraoxonase activity and SLE”
2008	Sangita Suresh	Ph.D.	“Functional characterization of <i>APOH</i> polymorphisms and their relation to SLE”
2009	Sarah Hill	M.S.	“Sequence variation in <i>APOA1</i> & <i>APOA4</i> and their relationship with plasma HDL-C levels”

e. Service on Comprehensive or Qualifying Examination Committees

Year Served	Student Population	Type of Exam
2006	1 Ph.D. Human Genetics student	Comprehensive
2007	1 Ph.D. Human Genetics student	Comprehensive

f. Other Teaching and Training

Year(s)	Teaching Activity	Description
<i>Training Undergraduate Students:</i>		
1999	Deniz Erturk	Summer Student
2006	Zahra Rahman	Summer Student
<i>Training Visiting Researchers:</i>		
2007	Fahad Waqar	MD - Visiting Researcher
2007-2008	Varinderjit Parmar	MD - Visiting Researcher
2007-2008	Ali Zaidi	MD - Visiting Researcher
2008	Mohammad Irfan	MD - Visiting Researcher
<i>Training Rotation Graduate Students:</i>		
2008	Heejung Kang	Ph.D. Student
2008	Suhas Krishnamurthy	Ph.D. Student

2. RESEARCH AND SCHOLARLY ACTIVITIESa. Grants and Contracts Received**Principal Investigator/Recipient:**

Years Inclusive	Grant and/or Contract Number and Title	Source
2000-2001	Fellowship Grant: Molecular Genetics of XL Cone-Rod Dystrophy	Fight For Sight
<i>Pending</i> 2009-2013	R01 AR057028: Molecular Genetics of BAT genes and SLE risk	NIH/NIAMS

Co-Investigator:

Years Inclusive	Grant and/or Contract Number and Title	Source
2007-2011	R01 HL088648: Prothrombin gene variation and the risk of SLE and CHD (<i>M.I. Kamboh, PI</i>)	NIH/NHLBI
2007-2011	R01 HL084613: Genetic determinants of HDL-cholesterol (<i>M.I. Kamboh, PI</i>)	NIH/NHLBI
2008-2012	R01 HL092397: Immune/Inflammation genomics and the risk of SLE and CHD (<i>M.I. Kamboh, PI</i>)	NIH/NHLBI
2009-2014	R01 AG030653: Search for the Alzheimer's Gene on Chromosome 10 (<i>M.I. Kamboh, PI</i>)	NIH/NIA

b. Invited Lectureships and Seminars Related to Your Research

Year	Title of Presentation	Venue
2006	“New perspectives on RNA processing”	University of Michigan, Kellogg Eye Center, Ann Arbor, MI
2007	“Genetics of SLE”	Dean's Junior Faculty Seminar Series, GSPH, University of Pittsburgh, Pgh, PA

c. Other Research and Training Activities - selected

Year	Position	Description of Activity
2002	Participant	Genetics Review Course, ACMG, Chicago, IL
2002	Participant	43 rd Annual Short Course in Medical and Experimental Mammalian Genetics, The Jackson Laboratory, Bar Harbor, ME
2003	Participant	Successful Strategies for Research Recruitment, University of Pittsburgh, Pittsburgh, PA
2006	Participant	Genetic Basis of Adult Medicine, Harvard Medical School, Boston, MA
2006	Participant	Quantitative PCR, Microarrays and Biological Validation, CHI, Providence, RI
2007	Participant	Expression Data Analysis Course, Affymetrix University, Santa Clara, CA

d. Publications – attached

3. SERVICE ACTIVITIES (PROFESSIONALLY RELATED)**a. University/School/Department**

Years	Committee	Position
2003-2005	International Services Advisory Committee, University of Pittsburgh	Member
2007-2009	Dean's Day, GSPH, University of Pittsburgh	Juror
2007-Present	GSPH Safety Committee, University of Pittsburgh	Member
2009-Present	Student Admissions Committee, Dept. of Human Genetics, GSPH, University of Pittsburgh	Member

b. Invited Reviewer - Manuscript and Other Document/Publication**Journal Title**

- Gene Therapy
- Ophthalmic Genetics
- American Journal of Ophthalmology
- Archives of Ophthalmology
- Molecular Vision
- Current Eye Research
- Ocular Immunology and Inflammation
- Human Immunology
- Journal of Biomedicine and Biotechnology

c. Study Sections, Review Panels, and Related Advisory Boards

Year	Position	Organization and Nature of Activity
2007	Grant Reviewer	University of Pittsburgh, Alzheimer Disease Research Center
2009	Grant Reviewer	NIH Challenge Grants

4. SERVICE ACTIVITIES (COMMUNITY RELATED)

Year	Position and Organization	Type of Service
2000	Johnston Elementary School	Health and education of the students during Health and Wellness Week

PUBLICATIONS

1. Refereed Articles (Published, In Press, Submitted)

1. **Demirci FY**, Dressen AS, Hamman RF, C. H. Bunker CH, Kammerer CM, Kamboh MI. Association of a common *G6PC2* variant with fasting plasma glucose levels in non-diabetic individuals (*submitted*)
2. Sanghera DK, **Demirci FY**, Been L, Ortega L, Ralhan SK, Wander GS, Mehra NK, Singh J, Aston CE, Mulvihill JJ, Kamboh MI. *PPARG* and *ADIPOQ* gene polymorphisms increase type 2 diabetes risk in Asian Indian Sikhs (*submitted*)
3. Heng C-K, He X, Saha N, Low PS, **Demirci FY**, Kamboh MI. Association of Three Lipoprotein Lipase Polymorphisms with Coronary Artery Disease in Chinese and Asian Indians. *International Journal of Cardiology* 2009 Jan 21. [Epub ahead of print]
4. Minster RL, **Demirci FY**, DeKosky ST, Kamboh MI. No association between *CALHMI* variation and risk of Alzheimer disease. *Human Mutation* 2009; 30:E566-569.
5. Suresh S, **Demirci FY**, Jacobs E, Kao AH, Rhew EY, Sanghera DK, Selzer F, Sutton-Tyrrell K, McPherson D, Bontempo FA, Kammerer CM, Ramsey-Goldman R, Manzi S, Kamboh MI. *APOH* Promoter Polymorphisms in Relation to Lupus and Lupus-Related Phenotypes. *Journal of Rheumatology* 2009; 36:315-322.
6. Figgins JA, Minster RL, **Demirci FY**, DeKosky ST, Kamboh MI. Association studies of 22 candidate SNPs with late-onset Alzheimer's disease. *Am J Med Genet B Neuropsychiatr Genet* 2009; 150B:520-526.
7. Taylor KE, Remmers EF, Lee AT, Ortmann WA, Plenge RM, Tian C, Chung SA, Nititham J, Hom G, Kao AH, **Demirci FY**, Kamboh MI, Petri M, Manzi S, Kastner DL, Seldin MF, Gregersen PK, Behrens TW, Criswell LA. Specificity of the STAT4 Genetic Association for Severe Disease Manifestations of Systemic Lupus Erythematosus. *PLoS Genetics* 2008; 4:e1000084.
8. Chen Q, Rhazzaghi H, **Demirci FY**, Kamboh MI. Functional significance of Lipoprotein Lipase *HindIII* polymorphism associated with the risk of coronary artery disease. *Atherosclerosis* 2008; 200:102-108.
9. Hom G, Graham RR, Modrek B, Taylor KE, Ortmann W, Garnier S, Lee AT, Chung SA, Ferreira RC, Pant K, Ballinger DG, Kosoy R, **Demirci FY**, Kamboh MI, Kao AH, Tian C, Gunnarsson I, Bengtsson AA, Rantapää-Dahlqvist S, Petri M, Manzi S, Seldin MF, Rönnblom L, Syvänen A-C, Criswell LA, Gregersen PK, Behrens TW. C8orf13/BLK and ITGAM/ITGAX are susceptibility loci for Systemic Lupus Erythematosus. *The New England Journal of Medicine* 2008; 358:900-909.
10. **Demirci FY**, Manzi S, Ramsey-Goldman R, Kenney M, Shaw PS, Dunlop-Thomas CM, Kao AH, Rhew EY, Bontempo F, Kammerer C, Kamboh MI. Association study of Toll-like

Receptor 5 (*TLR5*) and Toll-like Receptor 9 (*TLR9*) polymorphisms in Systemic Lupus Erythematosus. *Journal of Rheumatology* 2007; 34:1708-1711.

11. **Demirci FY**, Manzi S, Ramsey-Goldman R, Minster RL, Kenney M, Shaw PS, Dunlop-Thomas CM, Kao AH, Rhew E, Bontempo F, Kammerer C, Kamboh MI. Association of a common interferon regulatory factor 5 (*IRF5*) variant with increased risk of Systemic Lupus Erythematosus (SLE). *Annals of Human Genetics* 2007; 71:308-311.
12. **Demirci FY**, Chang M-H, Mah TS, Romero MF, Gorin MB. Proximal renal tubular acidosis and ocular pathology: a novel missense mutation in the gene (*SLC4A4*) for Sodium Bicarbonate Cotransporter protein (NBCe1). *Molecular Vision* 2006; 12:324-330.
13. **Demirci FY**, Rigatti BW, Mah TS, and Gorin MB. A novel *RPGR* exon ORF15 mutation in a family with X-linked retinitis pigmentosa and Coats-like exudative vasculopathy. *American journal of Ophthalmology* 2006; 141:208-210.
14. **Demirci FY**, Gupta N, Radak AL, Rigatti BW, Mah TS, Milam AH and Gorin MB. Histopathologic study of X-linked cone-rod dystrophy (*CORDX1*) caused by a mutation in the *RPGR* exon ORF15. *American journal of Ophthalmology* 2005; 139:386-388.
15. Gorin MB, Costantino J, Kulacoglu DN, **Demirci FY**, Wickerham DL, Fisher B and Wolmark N. Is tamoxifen a risk factor for retinal vaso-occlusive disease? *Retina* 2005; 25:523-526.
16. **Demirci FY**, Radak AL, Rigatti BW, Mah TS, and Gorin MB. A presumed missense mutation of *RPGR* causes abnormal RNA splicing with exon skipping. *American journal of Ophthalmology* 2004; 138:504-505.
17. **Demirci FY**, Rigatti BW, Mah TS, and Gorin MB. A novel compound heterozygous mutation in the cellular retinaldehyde-binding protein gene (*RLBPI*) in a patient with retinitis punctata albescens. *American journal of Ophthalmology* 2004; 138:171-173.
18. Jalkanen J, **Demirci FY**, Tyynismaa H, Bech-Hansen T, Meindl A, Peippo M, Mantyjarvi M, Gorin MB, and Alitalo T. A new genetic locus for X-linked progressive cone-rod dystrophy. *Journal of Medical Genetics* 2003; 40:418-423.
19. **Demirci FY**, Ramser J, White NJ, Rigatti BW, Meindl A, Lewis KF, Wen G, and Gorin MB. Refinement of the physical location and the genomic characterization of the CRSP2 (EXLM1) gene on Xp11.4. *DNA Sequence* 2003; 14:123-127.
20. Ayyagari R, **Demirci FY**, Liu J, Bingham EL, Stringham H, Kakuk LE, Boehnke M, Gorin MB, Richards JE, and Sieving PA. X-linked recessive atrophic macular degeneration from *RPGR* mutation. *Genomics* 2002; 80:166-171.
21. **Demirci FY**, Rigatti BW, Wen G, Radak AL, Mah TS, Baic CL, Traboulsi EI, Alitalo T, Ramser J, and Gorin MB. X-linked Cone-Rod Dystrophy (Locus *COD1*): Identification of mutations in *RPGR* exon ORF15. *American Journal of Human Genetics* 2002; 70:1049-1053.

22. Demirci H, Peksayar G, **Demirci FY**, Buyukbabani N, and Demiryont M. Bilateral microphthalmos with colobomatous orbital cyst. *Journal of Pediatric Ophthalmology and Strabismus* 2002; 39:110-113.
23. **Demirci FY**, White NJ, Rigatti BW, Lewis KF, and Gorin MB. Identification, genomic structure, and screening of the vacuolar proton-ATPase membrane sector-associated protein M8-9 gene within the COD1 critical region (Xp11.4). *Molecular Vision* 2001; 7:234-239.
24. Izgi B, Demirci H, **Demirci FY**, and Turker G. Diode laser cyclophotocoagulation in refractory glaucoma: comparison between pediatric and adult glaucomas. *Ophthalmic Surgery and Lasers* 2001; 32:100-107.
25. **Demirci FY**, Guney DB, Akarcay K, Kir N, Ozbek U, Sirma S, Unaltuna N, and Ongor E. Prevalence of factor V Leiden in patients with retinal vein occlusion. *Acta Ophthalmologica Scandinavica* 1999; 77:631-633.
26. **Demirci FY**, Kucukkaya R, Akarcay K, Kir N, Atamer T, Demirci H, and Ongor E. Ocular involvement in primary antiphospholipid syndrome. Ocular involvement in primary APS. *International Ophthalmology* 1998; 22:323-329.
27. **Demirci FY**, Sipahier A, and Turker G. [Glaucoma and optic disc]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1999; 29:180-187. Review.
28. Sezen F, Urgancioglu M, Tutkun IT, Demirci H, and **Demirci FY**. Comparative double-blind study of the efficacy of Lodoxamide 0.1% vs. N-acetyl aspartyl glutamic acid in patients with vernal keratoconjunctivitis. *Medical Bulletin of Istanbul Medical Faculty* 1999; 32:174-179.
29. Demirci H, Peksayar G, **Demirci FY**, and Yilmaz A. [Prognostic factors in retinoblastoma]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1999; 29:235-240.
30. Demirci H, **Demirci FY**, Turker G, and Izgi B. [Comparison of the short term effect of Metipranolol 0.1% with Timolol Maleat 0.5% and Betaxolol HCl 0.5% in the treatment of primary open angle glaucoma]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1998; 28:375-378.
31. Izgi B, Demirci H, **Demirci FY**, and Turker G. [Diode laser cyclophotocoagulation in the treatment of advanced refractory glaucoma]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1998; 28:18-25.
32. **Demirci FY**, Kucukkaya R, Akarcay K, Kir N, Atamer T, Demirci H, Ongor E, and Ovali T. [Ocular involvement in antiphospholipid syndrome]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1997; 27:278-284.
33. Sezen F, Gezer A, Demirci H, and **Karageyim FY**. [Long term results of phospholine iodide in convergence excess type accommodative esotropia]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1996; 26:244-248.
34. **Demirci FY**, Demirci H, and Bilgin LK. [Probing and irrigation in medical management of congenital nasolacrimal duct obstruction]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1995; 25:365-368.

35. Demirci H, **Demirci FY**, and Bilgin LK. [Hydroxyapatite orbital implant]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1995; 25:369-372.
36. Ovali T, Demirci H, Kir N, **Karageyim Y**, Gucukoglu A, and Basar D. [Choroidal detachment in retinal detachment surgery]. *Turkish Ophthalmology Journal (T. Oft. Gaz.)* 1994; 24:539-544.

2. Abstracts (Published, Submitted)

1. **Demirci FY**, Wang Y, Kao AH, Rhew EY, Ramsey-Goldman R, Manzi S, Kamboh MI. Comprehensive resequencing of F2 gene in Systemic Lupus Erythematosus. *Submitted for The American Society of Human Genetics 2009 Annual Meeting.*
2. Hill SE, **Demirci FY**, Dressen AS, Kammerer CM, Bunker CH, Hokanson JE, Hamman RF, Kamboh MI. Sequence variation in the APOA1 and APOA4 genes and their relationship with plasma HDL-cholesterol levels. *Submitted for The American Society of Human Genetics 2009 Annual Meeting.*
3. Burns LC, Minster RL, **Demirci FY**, DeKosky ST, Kamboh MI. Association studies of 12 candidate SNPs with late-onset Alzheimer's disease. *Submitted for The American Society of Human Genetics 2009 Annual Meeting.*
4. Kamboh MI, Hollister SM, Dressen AS, Bunker CH, Hamman RF, Kammerer CM, **Demirci FY**. *APOA2* genetic variation and its relationship with plasma HDL-cholesterol levels. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
5. **Demirci FY**, Dressen AS, Hamman RF, Kammerer CM, Kamboh MI. Association of a common *G6PC2* variant with fasting plasma glucose levels. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
6. Dasgupta S, **Demirci FY**, Dressen A, Kao A, Kammerer C, Manzi S, Kamboh MI. Relation of Paraoxonase (*PON2*) Polymorphisms with PON Activity and Systemic Lupus Erythematosus (SLE). *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
7. Suresh S, **Demirci FY**, Kao AH, Kammerer CM, Manzi S, Kamboh MI. Functional characterization of *APOH* promoter and its variants. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
8. Waqar F, **Demirci FY**, Dressen AS, Bunker CH, Hamman RF, Kammerer CM, Kamboh MI. *APOM* sequence analysis in relation to plasma HDL-C levels. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
9. Sanghera DK, **Demirci FY**, Ortega L, Been L, Ralhan SK, Wander GS, Mehra NK, Singh JR, Mulvihill JJ, Kamboh MI. Novel polymorphisms in PPARG and ADIPOQ genes increase type 2 diabetes risk in Asian Indian Sikhs: Evidence of gene-gene interaction. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.

10. Figgins JA, Minster RL, **Demirci FY**, DeKosky ST, Kamboh MI. Association Studies of 22 Candidate SNPs with Late-Onset Alzheimer's Disease. *The American Society of Human Genetics 2008 Annual Meeting*, November 11-15, 2008. Philadelphia, PA.
11. **Demirci FY**, Manzi S, Ramsey-Goldman R, Kao AH, Rhew EY, Bontempo F, Kammerer C, Kamboh MI. *TNFSF13 (APRIL)* polymorphisms and Systemic Lupus Erythematosus. *The American Society of Human Genetics 2007 Annual Meeting*, October 23-27, 2007. San Diego, CA.
12. Corthell LS, **Demirci FY**, Kao AH, Rhew EY, Bontempo F, Kammerer C, Ramsey-Goldman R, Manzi S, Kamboh MI. Relationship of Apolipoprotein H (*APOH*) polymorphisms with susceptibility to Systemic Lupus Erythematosus. *The American Society of Human Genetics 2007 Annual Meeting*, October 23-27, 2007. San Diego, CA.
13. Dasgupta S, **Demirci FY**, Kao AH, Rhew EY, Bontempo F, Kammerer C, Ramsey-Goldman R, Manzi S, Kamboh MI. *PON2* polymorphisms, PON activity, and Systemic Lupus Erythematosus (SLE). *The American Society of Human Genetics 2007 Annual Meeting*, October 23-27, 2007. San Diego, CA.
14. Suresh S, Jacobs E, Manzi S, Sanghera DK, Kao AH, Bontempo F, Kammerer C, **Demirci FY**, Kamboh MI. Association of *APOH* promoter polymorphisms with lupus nephritis and cardiovascular disease. *The American Society of Human Genetics 2007 Annual Meeting*, October 23-27, 2007. San Diego, CA.
15. **Demirci FY**, Manzi S, Minster RL, Bontempo F, Shaw P, Kao A, Kamboh MI. Association of a common interferon regulatory factor 5 (*IRF5*) variant with increased risk of Systemic Lupus Erythematosus (SLE). *The American Society of Human Genetics 2006 Annual Meeting*, October 9-13, 2006. New Orleans, LA
16. Dasgupta S, **Demirci FY**, Minster RL, Kenney M, Shaw P, Kao A, Kammerer C, Bontempo F, Manzi S, Kamboh MI. Association of *PON2* variants with PON activity in Systemic Lupus Erythematosus (SLE). *The American Society of Human Genetics 2006 Annual Meeting*, October 9-13, 2006. New Orleans, LA
17. Suresh S, Minster RL, **Demirci FY**, Kenney M, Shaw P, Kao A, Kammerer C, Bontempo F, Manzi S, Kamboh MI. Effect of *APOH* promoter SNPs on gene expression and plasma levels of APOH. *The American Society of Human Genetics 2006 Annual Meeting*, October 9-13, 2006. New Orleans, LA
18. **Demirci FY**, Rigatti BW, and Gorin MB. Human and mouse retinal alternative RNA splicing (AS) of a set of genes that are implicated in retinal degenerations. *The American Society of Human Genetics 2005 Annual Meeting*, October 25-29, 2005. Salt Lake City, UT
19. Gorin MB, **Demirci FY**, Chang M-H, Mah TS, Romero MF. A novel missense mutation in the Sodium Bicarbonate Cotransporter (*NBCe1/SLC4A4*) in a patient with proximal renal tubular acidosis and ocular pathology. *The American Society of Human Genetics 2005 Annual Meeting*, October 25-29, 2005. Salt Lake City, UT

20. **Demirci FY**, Rigatti BW, and Gorin MB. Retinal Alternative RNA Splicing of Murine Homologs of the Human Genes That Are Implicated in Retinal Degenerations. *ARVO 2005 Annual Meeting*, May 1-5, 2005. Fort Lauderdale, FL
21. Gorin MB, Rigatti BW, **Demirci FY**, Clarke SR, Mah TS, Weeks DE, and Ferrell RE. Refinement of the Cerulean cataract type 1 locus on 17q25.3 to a 0.5 Mb critical region. *The American Society of Human Genetics 2004 Annual Meeting*, October 26-30, 2004. Toronto, Canada
22. **Demirci FY**, Radak AL, Rigatti BW, Mah TS, and Gorin MB. A predicted missense mutation of *RPGR* causes aberrant RNA splicing with exon skipping. *Investigative Ophthalmology & Visual Science* 2004; 45:2485.
23. Kulacoglu DN, Costantino J, **Demirci FY**, Wickerham DL, Fisher B, Wolmark N, and Gorin MB. Tamoxifen and retinal vaso-occlusive disease. *Investigative Ophthalmology & Visual Science* 2004; 45:5269.
24. **Demirci FY**, Rigatti BW, Mah TS, and Gorin MB. A novel compound heterozygous mutation in the cellular retinaldehyde-binding protein gene (*RLBPI*) in a patient with retinitis punctata albescens. *American Journal of Human Genetics* 2003; 73(Suppl):2462.
25. Gorin MB, Rigatti BW, **Demirci FY**, Clarke SR, Mah TS, Weeks DE, Ferrell RE. Refinement and candidate gene screening of the Cerulean cataract type 1 locus on 17q24-q25. *American Journal of Human Genetics* 2003; 73(Suppl):1957.
26. **Demirci FY**, Radak AL, Rigatti BW, Traboulsi EI, Alitalo T, Mah TS, Gorin MB. Mutation screening of *RPGR* in male patients with X-linked or isolated forms of retinitis pigmentosa or cone-rod dystrophy. *Investigative Ophthalmology & Visual Science* 2003; 44:2312.
27. Chung DC, **Demirci YK**, Zegarra H, Locastro AJ, Gorin MB, Traboulsi E.I. Clinical correlation for two novel mutations in the *RPGR* gene for X-linked retinitis pigmentosa. *Investigative Ophthalmology & Visual Science* 2003; 44:1482.
28. Gorin MB, Rigatti BW, **Demirci FY**, Clarke SR, Mah TS, Radak AL, Weeks DE, Ferrell RE. Refinement and candidate gene screening of the Cerulean cataract type 1 locus on 17q24-q25. *Investigative Ophthalmology & Visual Science* 2003; 44:1260.
29. Radak AL, **Demirci FY**, Rigatti BW, Traboulsi EI, Alitalo T, Mah TS, Gorin MB. Mutation screening of *RPGR* in male patients with X-linked or sporadic forms of retinitis pigmentosa or cone-rod dystrophy. *American Journal of Human Genetics* 2002; 71(Suppl):2255.
30. **Demirci FY**, Radak AL, Rigatti BW, Mah TS, Gorin MB, Milam AH. Histopathology of a retina with X-linked cone-rod dystrophy caused by a mutation in the *RPGR* exon ORF15. *Investigative Ophthalmology & Visual Science* 2002; 43:2723.
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