CURRICULUM VITAE

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Academic Appointment:

Assistant Professor

Ophthalmic Research Center

Research Institute for Ophthalmology and Vision Science

Shahid Beheshti University of Medical Sciences

Address (work):

Ophthalmic Research Center

Shahid Beheshti University of Medical Sciences

No.23, Paidarfard St.

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Tehran, 16666, Iran.

Education:

1. 2001-2005 B.S. in Genetics, Shahid Chamran University

2. 2005-2008 MSc. in Cell and Molecular Biology, University of Tehran

Thesis defended in September 2008

Advisor: Professor Elahe Elahi

3. 2009-2015 Ph.D. in Cell and Molecular Biology, University of Tehran

Thesis defended in February 2015

Advisor: Professor Elahe Elahi

4. 2015-2016 Postdoctoral Fellowship, Shahid Beheshti University of Medical Sciences

Honors and Awards:

2010 Excellent Researcher Student Award, University of Tehran.

2012 Awarded by National Academy of Medical Sciences for second best publication in the basic sciences related to the medical sciences.

2013 Best article (in English), Shams Ophthalmology & Visual Sciences Festival.

2017 Best book chapter (translational), Shams Ophthalmology & Visual Sciences Festival.

2018 Excellent research award, 7th Iranian Research Association for Vision and Ophthalmology (IRAVO) congress.

2019 Excellent research award, 8h Iranian Research Association for Vision and Ophthalmology (IRAVO) congress.

Committee Assignments and Editorial Boards:

1. Member of the scientific committee, Annual congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)- Iranian Society of Ophthalmology (IrSO)
2. Member of prevention of inherited blinding eye diseases committee, Ministry of Health and Medical Education of Iran.
3. Member of Editorial Board, Journal of Ophthalmic and Vision Research.
4. Associate Editor, Journal of Gene, Cell and Tissue.
5. Reviewer, Journal of Ophthalmic and Vision Research.
6. Reviewer, Journal of Current Ophthalmology.

Major Research Interest

Genetics of human inherited ocular diseases, especially glaucoma and Inherited retinal dystrophies, Molecular mechanisms and novel therapeutic of eye diseases.

Research Grants:

As principal investigator (PI):

1. RPE65 gene screening in Iranian Leber Congenital Amaurosis (LCA) affected patients; Ophthalmic Research Center, Shahid Beheshti University of Medical Sciences; completed.
2. Histopathological investigation of fetus eye globe with identified homozygous mutation in CYP1B1 and predisposed to Primary Congenital Glaucoma (PCG), Shahid Beheshti University of Medical Sciences; completed.
3. Comprehensive genetic analysis of Iranian Leber Congenital Amaurosis (LCA) affected patients, Shahid Beheshti University of Medical Sciences; completed.
4. Comprehensive genetic analysis of Iranian stargardt affected patients, Shahid Beheshti University of Medical Sciences; completed.

5. Screening of 70 primary angle closure glaucoma (PACG) candidate genes in affected cohort by next generation sequencing (NGS), National Institute for Medical Research Development (NIMAD); in progress.
6. Screening of genes encoding MIR-200, MIR-146, MIR-126 and MIR-15 in sight-threatening diabetic retinopathy affected patients, Shahid Beheshti University of Medical Sciences; in progress.
7. Comparisons of expression of miRNAs affecting four major diabetic retinopathy (DR) pathogenic pathways in non-proliferative DR and proliferative DR patients with type 1 diabetes mellitus without DR, Shahid Beheshti University of Medical Sciences; in progress.
8. Search for causative genetic agent of aniridia in a large Iranian pedigree, Shahid Beheshti University of Medical Sciences; completed.
9. Comprehensive genetic analysis of Iranian keratoconus affected patients, Shahid Beheshti University of Medical Sciences; in progress.
10. TGFBI gene screening in Iranian TGFBI associated corneal dystrophies affected patients, Shahid Beheshti University of Medical Sciences; in progress.
11. Mutation screening of the bestrophin-1 gene in Iranian cohort with autosomal recessive bestrophinopathy and clinical investigation of the heterozygous carriers of the detected variants in the families, Shahid Beheshti University of Medical Sciences; in progress.

As co-investigator:

1. Investigation of role of LTBP2 in the TGFB2 signaling pathway and in response to oxidative stress. Ophthalmic Research Center, Shahid Beheshti University of Medical Sciences; completed.
2. Search for molecular mechanisms of pathogenesis affected by LTBT2 in glaucoma. Iran National Science Foundation (INSF); completed.
3. Pilot study on carrier frequency of common CYP1B1 mutations in Gilan province of Iran. Iran National Science Foundation (INSF); completed.
4. Search for novel glaucoma causing gene using linkage analysis and exome sequencing, Shahid Beheshti University of Medical Sciences; completed.
5. The Effect of Low Level Laser on Growth, Trans-Differentiation and De differentiation of RPE Cells in Adults versus Neonates; an in vitro study, Shahid Beheshti University of Medical Sciences; in progress.
6. Investigation on the probable role of cis p-tau in the pathogenesis of AMD, Shahid Beheshti University of Medical Sciences; in progress.
7. Study the effects of neutralizing antibody for Connective Tissue Growth Factor in reduction of choroidal neovascularization and its related scar tissue as intravitreal injection alone,

compared to combination with Avastin and Avastin only, in an animal model of laser induced Choroidal Neo-Vascularization (CNV), Shahid Beheshti University of Medical Sciences; completed.

8. Detecting the expression of NOGO-A and NOGO Receptor1 (NgR1) and LINGO1 in human RPE cell culture, Shahid Beheshti University of Medical Sciences; completed.
9. Estimation of carrier frequency of p.Arg368His mutation of CYP1B1 (primary congenital glaucoma causing gene) in the east of Gilan province, Shahid Beheshti University of Medical Sciences; completed.
10. Estimation of carrier frequency of p.Gly61Glu mutation of CYP1B1 (primary congenital glaucoma causing gene) in Talesh distinct of Gilan province, Shahid Beheshti University of Medical Sciences; completed.

International Publications (Articles):

1. Variable Expressivity and High Penetrance of CYP1B1 mutations associated with Primary Congenital Glaucoma. *Ophthalmology*, 2009; 116: 2101-2109.
2. Screening of Common CYP1B1 Mutations in Iranian POAG Patients using a Microarray-based PrASE Protocol. *Molecular Vision*, 2008; 14: 2349-2356.
3. Sex Bias in Primary Congenital Glaucoma Patients with and without CYP1B1 Mutations. *Journal of Ophthalmic and Vision Research*, 2009; 4: 75-78.
4. Loss of function mutations in the gene encoding latent transforming growth factor beta binding protein 2, LTBP2, cause primary congenital glaucoma. *Human Molecular Genetics*, 2009; 18: 3969-3977.
5. Contributions of MYOC and CYP1B1 mutations to JOAG. *Molecular vision*, 2008; 14:508-517.
6. Myocilin mutations are not a major cause of primary congenital glaucoma in Iranian patients. *Journal of ophthalmic and vision research*, 2010; 5: 101-104.
7. Contribution of the latent transforming growth factor-beta binding protein 2 gene to etiology of primary open angle glaucoma and pseudoexfoliation syndrome. *Molecular Vision*, 2013; 19: 333-347.
8. FOXC1 in human trabecular meshwork cells is involved in a regulatory pathway that includes miR-204, MEIS2 and ITG beta-1. *Experimental Eye Research*, 2013; 111: 112-121.
9. Diagnosis of cystathionine beta-synthase deficiency by genetic analysis. *Journal of Neurological Sciences*, 2014; 347: 305-309.
10. Glaucoma in Iran and contributions of studies in Iran to the understanding of the etiology of glaucoma. *Journal of ophthalmic and vision research*, 2015; 10(1): 68-76.

11. The p.Gly61Glu mutation in CYP1B1 affects the extracellular matrix in glaucoma patients. *Ophthalmic research*, 2016; 56(2): 98-103.
12. COL18A1 mutation identified as cause of iridocorneal angle closure: first identification of a causative gene for this major PACG. *Human Molecular Genetics*, 2018; 27(21): 3772-3786.
13. LTBP2 knockdown and oxidative stress affect glaucoma features including TGFβ pathways, ECM genes expression and apoptosis in trabecular meshwork cells. *Journal of Gene*, 2018; 673: 70-81.
14. P.Gly61Glu and p.Arg368His mutations in CYP1B1 that cause congenital glaucoma may be relatively frequent in certain regions of Gilan province, Iran. *Journal of ophthalmic and vision research*, 2018; 26 (2): 188-195.
15. Effects of intravitreal connective tissue growth factor neutralizing antibody on choroidal neovascular membrane-associated subretinal fibrosis. *Experimental eye research*, 2019; 184: 286-295.
16. A novel PAX6 mutation causes congenital aniridia with or without retinal detachment. *Ophthalmic Genetics*, 2019; 40(2): 146-149.
17. Incomplete penetrance of CRX gene for autosomal dominant form of cone-rod dystrophy. *Ophthalmic Genetics*, 2019; 40(3): 259-266.
18. PRPH2 mutation as the cause of various clinical manifestations in a family affected with inherited retinal dystrophy. *Ophthalmic Genetics*, 2019; 16: 1-7.
19. The First Inherited Retinal Disease Registry in Iran: Research Protocol and Results of a Pilot Study. *Archive of Iranian Medicine*, July 2020; 23(7): 445-454.
20. Association of Saitohin gene rs62063857 polymorphism with dry type age-related macular degeneration. *Ophthalmic Genetics*, 2020; 41(5): 505-506.
21. SVEP1 as a Genetic Modifer of TEK-Related Primary Congenital Glaucoma. *Investigative Ophthalmology & Visual Science*, 2020; 61(12): 6.
22. Choroidal Thickness in Different Types of Inherited Retinal Dystrophies. *Journal of ophthalmic and vision research*, 2020; 15 (3): 351-361.
23. Autosomal Recessive Bestrophinopathy: Clinical and Genetic Characteristics of Twenty-Four Cases. *Journal of Ophthalmology*, 2021, Article ID 6674290.
24. Variable Expressivity of Wolfram Syndrome in a Family with Multiple Affected Subjects. *Journal of ophthalmic and vision research*, 2021; 16 (4): 602-610.
25. Carrier Status for p.Gly61Glu and p.Arg368His CYP1B1 Mutations Causing Primary Congenital Glaucoma in Iran. *Journal of ophthalmic and vision research*, 2021; 16(4): 574-581.

26. Retinal Vascular Abnormalities in Different Types of Inherited Retinal Dystrophies Assessed by Optical Coherence Tomography Angiography. *Journal of Current Ophthalmology*, 2021; 33: 189-96.
27. Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. *Human Genetics*, 2022; 141(3-4): 785-803.
28. Intravitreal connective tissue growth factor neutralizing antibody or bevacizumab alone or in combination for prevention of proliferative vitreoretinopathy in an experimental model. *Experimental Eye Research*, 2021; 208: Article ID 108622.
29. Prenatal diagnosis of primary congenital glaucoma and histopathological features in a fetal globe with cytochrome p4501B1 mutations. *European Journal of Ophthalmology*, 2021; 3: Article ID 11206721211051235.
30. Mutation Screening of Six Exons of ABCA4 in Iranian Stargardt Disease Patients. *Journal of ophthalmic and vision research*, 2022; 17 (1): 51-58.
31. Identification of three novel homozygous variants in COL9A3 causing autosomal recessive Stickler syndrome. *Orphanet Journal of Rare Diseases*, 2022; 17: 97.
32. Genetic screening of TGFBI in Iranian patients with TGFBI-associated corneal dystrophies and a metaanalysis of global variation frequencies. *Ophthalmic Genetics*, 2022; 43(4): 496-499.
33. Augmented Expression of NOGO-A and Its Receptors in Human Retinal Pigment Epithelial Cells Following Treatment with Human Amniotic Fluid. *Iranian Journal of Public Health*, 2022; 51(7): 1658-1666.
34. The Inhibitory Effect of Connective Tissue Growth Factor Antibody on Postoperative Fibrosis in a Rabbit Model of Trabeculectomy. *Journal of ophthalmic and vision research*, 2022; 17(4): 486-496.
35. Clinical and economic analysis of the policy to prevent the birth of Down syndrome fetuses and evaluation of clinical ophthalmic manifestations in affected individuals. *Sarem Journal of Medical Research*, 2022; 7(1): 29-39.
36. Choroidal structure investigated by choroidal vascularity index in patients with inherited retinal diseases. *International Journal of Retina and Vitreous*, 2023; 9: 18.
37. A health terminological system for inherited retinal diseases: Content coverage evaluation and a proposed classification. *PLoS ONE*, 18(8): e0281858.
38. Expression Profiling of ADAMTS (L) Superfamily of Genes in Various Human Eye Tissues. *Iranian Journal of Public Health*, 2023; 52(12): 2630-2642.

International Conferences:

1. Incomplete penetrance of G61E and R390H mutations in CYP1B1 among Iranian primary congenital glaucoma patients. *European journal of Human Genetics (ESHG 2007)*, volume 15, supplement 1, June 2007.
2. Discordance of primary congenital glaucoma in monozygotic twins. *European journal of Human Genetics (ESHG 2008)*, volume 16, supplement 2, May 2008.
3. One genotype-six different phenotype: variable expression not incomplete penetrance. *15th national and 3rd international conference of Biology*, University of Tehran, Tehran, Iran, 19-21 August, 2008.
4. Using multi-sample slides spotted with universal probes to detect common CYP1B1 mutations in primary open angle glaucoma patients. *The 9th Iranian congress of Biochemistry & the 2nd International congress of Biochemistry and Molecular Biology*, Shiraz, Iran, Oct, 29- Nov, 1, 2007.
5. Non-disease associated variations of CYP1B1 and disease associated variations with incomplete penetrance are mostly located in loop regions of the CYP1B1 protein. *Bioinformatics congress*, IBB, university of Tehran, Tehran, Iran, 2008.
6. Hybridization of multiplex PrASE products to oligonucleotide spotted microarrays. *The 9th Iranian congress of Biochemistry & the 2nd International congress of Biochemistry and Molecular Biology*, Shiraz, Iran. Oct, 29- Nov, 1, 2007.
7. Role of CYP1B1 mutations in Iranian POAG patients assessed by Microarray based PrASE protocol. *Asia ARVO, International Meeting on Research in vision and Ophthalmology*, Hyderabad, India, January 15-18, 2009.
8. Absence of linkage to GLC3B and GLC3C in Iranian Primary Congenital Glaucoma (PCG) pedigrees without mutations in CYP1B1 and MYOC. *Asia ARVO, International Meeting on Research in vision and Ophthalmology*, Hyderabad, India, January 15-18, 2009.
9. The effect of ice water stress and chronic administration of estradiol benzoate on acute pain in ovarectomized albino mice. *16th Iranian congress of physiology & pharmacology*, Tehran, Iran, May 9-13 2003.
10. Homozygosity mapping in one Iranian pedigree affected with Primary Congenital Glaucoma (PCG) reveals linkage to GLC3B locus. *Basic and clinical neuroscience congress*, 2012, Tehran, Iran. 7-9 Nov 2012.
11. Contribution of LTBP2 with etiology of Primary Angle Closure Glaucoma. *Basic and clinical neuroscience congress*, 2012, Tehran, Iran. 7-9 Nov 2012.
12. A frameshift mutation in cytochrome P4501B1 (CYP1B1) as the principal cause of primary congenital glaucoma (PCG) in a family linked to the GLC3A locus. *The 3rd*

Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO), Tehran, Iran, March 7-8, 2013.

13. Contribution of LTBP2 to etiology of primary open angle glaucoma and pseudoexfoliation syndrome. *The 3rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, March 7-8, 2013.
14. Diagnosis of homocystinuria in Anterior Segment Dysgenesis (ASD) pedigree by whole genome genetic analysis. *The 4rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, February 13, 2014.
15. FOXC1 regulates expression of CLOCK, GNG5, CXCL6, ITGβ1, LDLRAD2, FMNL2, KHDRBS3, MEIS2, PLEKHG5, and WWC2 in human trabecular meshwork cells. *The 4rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, February 13, 2014.
16. Dexamethasone Treatment Reduces LTBP1 Expression in Human Trabecular Meshwork Cultured Cells. *The 4rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, February 13, 2014.
17. Homozygosity mapping used for identification of disease causing genes in heterogenic disorders. *1st international & 13th Iranian Genetic congress*, Tehran, Iran, May 24-26, 2014.
18. Whole exome sequencing analysis in a large Primary Angle Closure Glaucoma (PACG) pedigree. *European journal of Human Genetics (ESHG 2014)*, volume 22, supplement 1, 31 May- 3 June, 2014.
19. Role of LTBP2 in Glaucoma Pathogenesis. *The 5rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, March 5-6, 2015.
20. MIR184 mutations not observed in 47 Iranian Keratoconus patients. *The 5rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, March 5-6, 2015.
21. Glaucomatous risk factors and trabecular meshwork extracellular matrix remodeling. *The 6rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, March 3-4, 2016.
22. Oxidative stress effects in glaucoma may be mediated by the TGF-β signaling pathway. *The Association for Research in Vision and Ophthalmology (ARVO)*, Seattle, Washington, United States, May 1-5, 2016. Investigative Ophthalmology & Visual Science 57 (12), 6015-6015.

23. NOGO-A Gene Expression in Amniotic Fluid Treated Human RPE Cells. *The Association for Research in Vision and Ophthalmology (ARVO)*, Baltimore, MD, United States, May 7-11, 2017. Investigative Ophthalmology & Visual Science 58 (8), 608-608.
24. Investigation of role of some glaucoma risk factors in the disease pathogenesis. *European Society of Human Genetics (ESHG)*, 2017.
25. COL18A1 mutation identified as cause of iridocorneal angle closure: first identification of a causative gene for this major PACG presentation. *The 7rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2017.
26. Risk of angle closure glaucoma for carriers of Knobloch syndrome. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
27. Two novel mutations in CHM identified as cause of choroidemia in two unrelated Iranian affected families. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
28. PAX6 gene screening in a large Iranian pedigree with aniridia. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
29. Possible bidirectional regulation of PITX2 and TGF-beta signaling in human trabecular meshwork cells. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
30. Micro-RNAs that target genes potentially related to glaucoma. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
31. Estimation of frequencies of four common primary congenital glaucoma causing mutations in CYP1B1 in the province of Gilan. *The 8rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO)*, Tehran, Iran, April, 2018.
32. Enhanced Expression of NOGO-A Gene and its Receptors in Amniotic Fluid Treated Human RPE Cells. *The Association for Research in Vision and Ophthalmology (ARVO)*, Hawaii Convention Center Honolulu, Hawaii, United States, April 29 – May 3, 2018. Investigative Ophthalmology & Visual Science 59 (9), 3087-3087.
33. Intravitreal Connective Tissue Growth Factor Neutralizing Antibody Reduces Subretinal Fibrosis Associated with Experimental Choroidal Neovascular Membrane. *The Association for Research in Vision and Ophthalmology (ARVO)*, Vancouver

Convention Centre, Vancouver, B.C. Canada, April 28 – May 2, 2019. Investigative Ophthalmology & Visual Science 60 (9), 2978-2978.

34. Molecular Analysis of twenty-seven Iranian Patients with Usher Syndrome. *4th International Symposium on Usher syndrome,* Mainz, Germany, 2018.
35. Exome sequencing uncovers multilocus genomic variation and unexpected molecular heterogenety in patients with retinal degeneration and hearing impairment. *German Society of Human Genetics,* Annual meeting Weimar, 2019.
36. Genetic screening of Twenty-four Iranian Families Affected with Retinitis Pigmentosa. *The 9rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO),* Tehran, Iran, April 18-19, 2019.
37. Identification of mutation in PRPH2 gene as cause of disease in a complex family affected with inherited retinal dystrophies by exome sequencing. *The 9rd Annual Congress of Iranian Research Association for Vision and Ophthalmology (IRAVO),* Tehran, Iran, April 18-19, 2019.
38. Angiopoietin Receptor TEK-Associated Primary Congenital Glaucoma: Five Novel Gene Variants with Phenotype Expansion. *8th world glaucoma congress,* Melbourne, England, March 27-30, 2019.