

MAJID FARDAEI

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Professional Summary

A Molecular Geneticists, who has performed different techniques during M.Sc., PhD and Post-Doctoral followed by training of M.Sc. and PhD students. During my PhD I identified and cloned two new human genes which are involved in Myotonic Dystrophy. Also I have been a technical officer in genetic diagnostic laboratory for 7 years in which different molecular genetic methods for diagnosis of genetic abnormality has been used. As a result I identified and reported new mutations in different genetic disorders. Recently in collaboration with Rabinowitz YS in UCLA, USA we identified a new gene which is involved in Keratoconus Patients. I also designed and produced a novel T-vector plasmid construct that simplifies gene cloning, and a QF-PCR based diagnostic kit for detection of chromosome 21, 13, 18, X and Y aneuploidies using 26 microsatellite and STS markers in a single multiplex PCR. Further to above mentioned activities, I have set up and managed a molecular genetics laboratory in South of Iran.

Experiences

- Bacterial culture
- Yeast three hybrid system
- Recombinant DNA technology
- Gene Synthesis
- PCR
- Genetic Analyzer
- Sequencing
- QF-PCR
- MLPA
- Western Blotting
- HPLC
- Tissue culture techniques
- Plasmid and DNA purification
- Plasmid Design and construction
- CRISPR gene editing
- RT-PCR
- Bioinformatics: Primer design software, Sequence analysis, AlleleID, ClinGen, UCSC, Ensemble, Varsome, Mastermind, NCBI Tools, COSMIC, Sequence analysis tools, AlleleID, and many more.

Activity

- Member of editorial review board of Iranian Journal of Medical Sciences (<http://ijms.sums.ac.ir/index.php/IJMS/pages/view/EditorialTeam>)
- Teaching and supervising PhD students (three students)
Student names and Thesis subjects:
1-Student Name: Fallahi J.
Subject: Molecular analysis of recurrent hidatidiform mole and functional analysis of normal and mutant variants of KHDC3L gene in genomic methylation. 2015-2019

2-Student Name: Miri M. R.
Subject: QF-PCR based diagnostic kit for detection of chromosome 21 and 18 aneuploidies using microsatellite and STS markers in a single multiplex PCR. 2014-2018

3-Student Name: Saberzadeh J.
Subject: QF-PCR based diagnostic kit for detection of chromosome 13, X and Y aneuploidies using microsatellite and STS markers in a single multiplex PCR. 2014-2018
- Teaching and supervising M.Sc. students (ten students)
Three sample Thesis subjects:
1-Student Name: Sadeghipour F.
Subject: Mutation analysis of the CTNS gene in Iranian patients with infantile nephropathic cystinosis. 2014-2016

2-Student Name: Rezaei M.
Subject: Genetic analysis of patients with recurrent hydatidiform mole. 2013-2015

3-Student Name: Dastsooz H.
Subject: ATP7B gene mutation analysis in Iranian families with Wilson disease. 2010-2012
- Teaching medical genetics to undergraduate students: Medical, Dentistry, Nursing, Midwifery, and Laboratory Sciences.
- Patenting and marketing QF-PCR based diagnostic kit for detection of chromosome 21, 13, 18, X and Y aneuploidies using 26 microsatellite and STS markers in a single multiplex PCR.

Employment

2006-Present: Associate Professor, Shiraz University of Medical Sciences, Shiraz, Iran,

2002-2006: Post Doctoral Research Fellow, Nottingham University, UK. Subject: Using A New Site Specific Recombinase System in Embryonic stem cells.

Education

Ph.D., Genetics: Genetics Department, Nottingham, UK, Theses: Molecular Mechanism of Myotonic Dystrophy.

M.Sc., Biochemistry: Biochemistry Department, Shiraz University of Medical Sciences, Shiraz, Iran.

B.Sc., Biology: Biology Department, Shahid Chamran University, Ahvaz, Iran.

Award

- The best presentation in Second International Congress of Molecular Biology, 2007.

Publication

1. Commentary: Profiling of UGT1A1*6, UGT1A1*60, UGT1A1*93, and UGT1A1*28 Polymorphisms in Indonesian Neonates With Hyperbilirubinemia Using Multiplex PCR Sequencing. Heydari MR, **Fardaei M**. Front Pediatr. 2020 Feb 7;7:528.
2. Association of FTO rs9939609 polymorphism with serum leptin, insulin, adiponectin, and lipid profile in overweight adults. Mehrdad M, Doaei S, Gholamalizadeh M, **Fardaei M**, Fararouei M, Eftekhari MH. Adipocyte. 2020 Dec;9(1):51-56.
3. Two novel mutations in the MECP2 gene in patients with Rett syndrome. Khalili Alashti S, Fallahi J, Mohammadi S, Dehghanian F, Farbood Z, Masoudi M, Poorang S, Jokar A, **Fardaei M**. Gene. 2020 Jan 17;732:144337. doi: 10.1016/j.gene.2020.144337.
4. Further delineation of the phenotype caused by a novel large homozygous deletion of GRID2 gene in an adult patient. Taghdiri M, Kashef A, Abbassi G, Moshtagh A, Sadatian N, **Fardaei M**, Najafi K, Kariminejad R. Clin Case Rep. 2019 May 4;7(6):1149-1153. doi: 10.1002/ccr3.2020. eCollection 2019 Jun.

5. Segmental Duplications as a Complement Strategy to Short Tandem Repeats in the Prenatal Diagnosis of Down Syndrome. Miri MR, Saberzadeh J, Behzad Behbahani A, Tabei MB, Alipour M, **Fardaei M**. Iran J Med Sci. 2019 May;44(3):214-219.
6. A Novel Mutation in NLRP7 Related to Recurrent Hydatidiform Mole and Reproductive Failure. Fallahi J, Razban V, Momtahan M, Akbarzadeh-Jahromi M, Namavar-Jahromi B, Anvar Z, **Fardaei M**. Int J Fertil Steril. 2019 Jul;13(2):135-138. doi: 10.22074/ijfs.2019.5657. Epub 2019 Apr 27.
7. An immunocompetent patient with a nonsense mutation in NHEJ1 gene. Esmaeilzadeh H, Bordbar MR, Hojaji Z, Habibzadeh P, Afshinfar D, Miryounesi M, **Fardaei M**, Faghihi MA. BMC Med Genet. 2019 Mar 21;20(1):45. doi: 10.1186/s12881-019-0784-0.
8. Novel mutation in the *MED23* gene for intellectual disability: A case report and literature review. Hashemi-Gorji F, **Fardaei M**, Tabei SMB, Miryounesi M. Clin Case Rep. 2019 Jan 9;7(2):331-335. doi: 10.1002/ccr3.1942. eCollection 2019 Feb.
9. Enhancing Stability of Destabilized Green Fluorescent Protein Using Chimeric mRNA Containing Human Beta-Globin 5' and 3' Untranslated Regions. Adibzadeh S, **Fardaei M**, Takhsid MA, Miri MR, Rafiei Dehbidi G, Farhadi A, Ranjbaran R, Alavi P, Nikouyan N, Seyyedi N, Naderi S, Eskandari A, Behzad-Behbahani A. Avicenna J Med Biotechnol. 2019 Jan-Mar;11(1):112-117.
10. The First Case of a Small Supernumerary Marker Chromosome 18 in a Klinefelter Fetus: A Case Report. Saberzadeh J, Miri MR, Dianatpour M, Behzad Behbahani A, Tabei MB, Alipour M, Faghihi MA, **Fardaei M**. Iran J Med Sci. 2019 Jan;44(1):65-69.
11. Advanced molecular approaches pave the road to a clear-cut diagnosis of hereditary retinal dystrophies. Ravesh Z, Dianatpour M, **Fardaei M**, Taghdiri M, Hashemi-Gorji F, Yassae VR, Miryounesi M. Mol Vis. 2018 Oct 19;24:679-689. eCollection 2018.
12. The genetics of recurrent hydatidiform moles: new insights and lessons from a comprehensive analysis of 113 patients. Nguyen NMP, Khawajkie Y, Mechtouf N, Rezaei M, Breguet M, Kurvinen E, Jagadeesh S, Solmaz AE, Aguinaga M, Hemida R, Harma MI, Rittore C, Rahimi K, Arseneau J, Hovanes K, Clisham R, Lenzi T, Scurry B, Addor MC, Bagga R, Nendaz GG, Finci V, Poke G, Grimes L, Gregersen N, York K, Bolze PA, Patel C, Mozdarani H, Puechberty J, Scotchie J, **Fardaei M**, Harma M, Gardner RJM, Sahoo T, Dudding-Byth T, Srinivasan R, Sauthier P, Slim R.
13. Clinical and genetic-epigenetic aspects of recurrent hydatidiform mole: A review of literature. Moein-Vaziri N, Fallahi J, Namavar-Jahromi B, **Fardaei M**, Momtahan M, Anvar Z. Taiwan J Obstet Gynecol. 2018 Feb;57(1):1-6. doi: 10.1016/j.tjog.2017.12.001. Review.
14. Identification of a novel deletion within *ALDH3A2* gene in an Iranian Family with Sjögren-Larsson Syndrome. Taghdiri M, Kashef A, **Fardaei M**, Miryounesi M. Clin Case Rep. 2017 Nov 22;6(1):32-36. doi: 10.1002/ccr3.1235. eCollection 2018 Jan.
15. Does the c.-273T>C variant in the upstream region of the *HBB* gene cause a thalassemia phenotype? Dastsooz H, Alipour M, Mohammadi S, Dehghanian F, Kamgarpour F, **Fardaei M**. Blood Res. 2017 Dec;52(4):332-334. doi: 10.5045/br.2017.52.4.332. Epub 2017 Dec 26. No abstract available.
16. TSC1 Mutations in Keratoconus Patients With or Without Tuberous Sclerosis. Bykhovskaya Y, **Fardaei M**, Khaled ML, Nejabat M, Salouti R, Dastsooz H, Liu Y, Inaloo S, Rabinowitz YS. Invest Ophthalmol Vis Sci. 2017 Dec 1;58(14):6462-6469. doi: 10.1167/iovs.17-22819.
17. Mutation analysis of the *CTNS* gene in Iranian patients with infantile nephropathic cystinosis: identification of two novel mutations. Sadeghipour F, Basiratnia M, Derakhshan A, **Fardaei M**. Hum Genome Var. 2017 Oct 5;4:17038. doi: 10.1038/hgv.2017.38. eCollection 2017.
18. Limb Girdle Muscular Dystrophy Type 2E Due to a Novel Large Deletion in SGCB Gene. Ghafouri-Fard S, Hashemi-Gorji F, **Fardaei M**, Miryounesi M. Iran J Child Neurol. 2017 Summer;11(3):57-60.
19. A Novel Mutation in *ERCC8* Gene Causing Cockayne Syndrome. Taghdiri M, Dastsooz H, **Fardaei M**, Mohammadi S, Farazi Fard MA, Faghihi MA. Front Pediatr. 2017 Aug 9;5:169. doi: 10.3389/fped.2017.00169. eCollection 2017.
20. Novel mutations in PANK2 and PLA2G6 genes in patients with neurodegenerative disorders: two case reports. Dastsooz H, Nemat H, Fard MAF, **Fardaei M**, Faghihi MA. BMC Med Genet. 2017 Aug 18;18(1):87. doi: 10.1186/s12881-017-0439-y.

21. *VSX1* and *SOD1* Mutation Screening in Patients with Keratoconus in the South of Iran. Nejabat M, Naghash P, Dastsooz H, Mohammadi S, Alipour M, **Fardaei M**. J Ophthalmic Vis Res. 2017 Apr-Jun;12(2):135-140. doi: 10.4103/jovr.jovr_97_16.
22. Linkage Analysis based on Four Microsatellite Markers to Screen for Unknown Mutation in Families with Wilson Disease. Arianfar F, **Fardaei M**. Clin Lab. 2016 Aug 1;62(8):1541-1546. doi: 10.7754/Clin.Lab.2016.160109.
23. Second Allele Finder Software: a Simple Approach Toward HLA Typing. Fadaie Z, Ahmadzadeh M, Salehi M, Bordbar M, **Fardaei M**. Clin Lab. 2016 Oct 1;62(10):2045-2051. doi: 10.7754/Clin.Lab.2016.160220.
24. Expression Analysis of Multiple Genes May Involve in Antimony Resistance among *Leishmania major* Clinical Isolates from Fars Province, Central Iran. Ghobakhloo N, Motazedian MH, **Fardaei M**. Iran J Parasitol. 2016 Apr-Jun;11(2):168-176.
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27. Two novel mutations in the *KHDC3L* gene in Asian patients with recurrent hydatidiform mole. Rezaei M, Nguyen NM, Foroughinia L, Dash P, Ahmadpour F, Verma IC, Slim R, **Fardaei M**. Hum Genome Var. 2016 Sep 1;3:16027. doi: 10.1038/hgv.2016.27. eCollection 2016.
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31. The genomic architecture of NLRP7 is Alu rich and predisposes to disease-associated large deletions. Reddy R, Nguyen NM, Sarabay G, Rezaei M, Rivas MC, Kavasoglu A, Berkil H, Elshafey A, Abdalla E, Nunez KP, Dreyfus H, Philippe M, Hadipour Z, Durmaz A, Eaton EE, Schubert B, Ulker V, Hadipour F, Ahmadpour F, Touitou I, **Fardaei M**, Slim R. Eur J Hum Genet. 2016 Oct;24(10):1445-52. doi: 10.1038/ejhg.2016.9. Epub 2016 Mar 9. Erratum in: Eur J Hum Genet. 2016 Oct;24(10):1516.
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33. Impact of *KIF6* Polymorphism rs20455 on Coronary Heart Disease Risk and Effectiveness of Statin Therapy in 100 Patients from Southern Iran. Hamidizadeh L, Haji Hosseini Baghdad Abadi R, Babaei Baigi MA, Dastsooz H, Khazaei Nejhad A, **Fardaei M**. Arch Iran Med. 2015 Oct;18(10):683-7. doi: 0151810/AIM.008.
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38. A new missense mutation in the BCKDHB gene causes the classic form of maple syrup urine disease (MSUD). Miryounesi M, Ghafouri-Fard S, Goodarzi H, **Fardaei** M. *J Pediatr Endocrinol Metab*. 2015 May;28(5-6):673-5. doi: 10.1515/jpem-2014-0341.
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42. MSX1 mutation in witkop syndrome; a case report. Ghaderi F, Hekmat S, Ghaderi R, **Fardaei** M. *Iran J Med Sci*. 2013 Jun;38(2 Suppl):191-4.
43. Multiplex ARMS PCR to Detect 8 Common Mutations of ATP7B Gene in Patients With Wilson Disease. Dastsooz H, Imanieh MH, Dehghani SM, Haghighat M, Moini M, **Fardaei** M. *Hepat Mon*. 2013 May 16;13(5):e8375. doi: 10.5812/hepatmon.8375. eCollection 2013.
44. A new ATP7B gene mutation with severe condition in two unrelated Iranian families with Wilson disease. Dastsooz H, Dehghani SM, Imanieh MH, Haghighat M, Moini M, **Fardaei** M. *Gene*. 2013 Feb 1;514(1):48-53. doi:10.1016/j.gene.2012.10.085. Epub 2012 Nov 13.
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46. Expression of $\alpha 2$, $\alpha 5$ and $\alpha 6$ subunits of integrin in de-differentiated NIH3T3 cells by cell-free extract of embryonic stem cells. Mostafavi-Pour Z, Keihani S, Talaei-Khozani T, Mokaram P, **Fardaei** M, Rohani L, Ebadat S, Sardarian A. *Mol Biol Rep*. 2012 Jul;39(7):7339-46. doi: 10.1007/s11033-012-1565-4.
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50. In vivo co-localisation of MBNL protein with DMPK expanded-repeat transcripts. **Fardaei** M, Larkin K, Brook JD, Hamshire MG. *Nucleic Acids Res*. 2001 Jul 1;29(13):2766-71.