Curriculum Vitae

Dr Mahmoud Shekari Khaniani, M.D, Ph.D

Associate Professor in Human Genetics

Personal details:

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Education:

2004 - 2007: **Human Genetics, Doctorate (PhD)**

Paediatric Department, Faculty of Medicine, Dentistry and Health Sciences, Cyto-Molecular Research Group, Murdoch Children research Institute, Royal children's Hospital, University of Melbourne, Australia

Thesis Title: Molecular characterization and assessment of clinical significance of small Fragile X alleles

Supervisor: Professor Andy Choo and A/Pro Howard Slater

1987-1994: **Medical Doctor (M.D)**

Faculty of Medicine, Tabriz University of Medical science, Tabriz, Iran

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Clinical and work Experience:

- **A**ssociate Professor in Human Genetics at the Department of Medical Genetics, Tabriz university of Medical Science (2008 so far)

- Director of Ebna cina Medical Genetic Diagnostic Centre, Specialized and Sub specialized Outpatient Clinic of Tabriz University of Medical Sciences, No 507 ,Sheikh Alraeis Azadi Ave, Tabriz, Tran
- Clinical Genetic Consoler in Specialized and Sub specialized Outpatient Clinic of Tabriz University of Medical Sciences (2008 so far)
- Working in hospitals as a General Practitioner
- Working in health centres as a Family Physician

Research Experience:

- **Supervised over 35 MSc and Ph.D. Thesis's**
- Expression of cDNA human F8 gene from Recombinant DNA clone in hamster ovary cells (2010 - in progress)
- Production of Human Epidermal Growth Factor (EGF) from Recombinat DNA clone in Escherichia coli and Yeast (2010 - in progress)
- Production of Vascular Epidermal Growth Factor (VEGF) from Recombinat DNA clone in Escherichia coli and Yeast (2012 - in progress)
- Production of Human Stem Cell Factor (SCF) from Recombinat DNA clone in Escherichia coli and Yeast (2010- in progress)
- Molecular characterization and assessment of clinical significance of small Fragile X alleles (2004-2006)
- Gene induction for the treatment of Methylmalonic acidouria(2002-2004)

Experimental Techniques:

Molecular and cell biology

- Routine bacterial cell culture technique (bacterial cultivation, preparation and transformation of competent cells)
- Routine DNA manipulation techniques (cloning, plasmid and bacterial artificial chromosome construction, transfection).
- DNA, RNA extraction from bacterial and mammalian cells and tissues
- Polymerase chain reaction, PCR, (amplification including long range and screening), sequencing, revers transcription and real-time PCR
- Manipulation of large BACs by purification, digestion and transfection)
- PFGE and agars gel electrophoresis (DNA)

Tissue Culture

- Culture of lymphocytes
- Culture of Amniotic fluid

- Routine cell culture and passage
- Freezing stocks of mammalian cells
- Thawing of frozen mammalian cells
- Primary culturing mammalian cells (fibroblast, kidney, brain, liver…)
- Maintenances of cell line cultures
- Counting cells and calculation of cell size using hemocytometer
- DNA transfection into human cells and produce stable cell line
- Microscopy (phase contrast, fluorescent and confocal)
- Media preparation
- Experience with tissue collection and biopsies (human and mouse)
- Flow cytometry
- Isolation of liver stem cells from mouse embryo
- Isolation of heamopotic stem cells from adult mouse

Communication

1. Knowledge of English language, Reading, Writing, speaking and listening
2. Preparation of scientific poster presentation and paper
3. Presentations at laboratory meetings and journal clubs
4. Familiarity with computer programs such as word, PowerPoint, Excel, experienced in database searches

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Teaching experiences

- Medical Genetics for medical students
- Medical Genetics for dentistry students
- Human Cryogenetics for Msc student
- Medical Genetics for Msc and Ph.D student
- Molecular Genetics for Msc and Ph.D student
- Genetic engineering for Msc and Ph.D student
- Advanced Molecular Genetics for Msc and Ph.D student
- Medical Genetics Course for Medical and dentistry residency student

Publication:

1. **Mahmoud, S Khaniani** , Paul Kalitsis, Trent Burgess, Howard R Slater
   An Improved Diagnostic PCR Assay for Identification of Cryptic Heterozygosity for CGG Triplet Repeat Alleles in the Fragile X Gene (FMR1). Journal of ***Molecular Cytogenetics*** 2008; 1:5 Page 1-6
2. Ruimei Hu, Nicole E. Buck, **Mahmoud S. Khaniani**, Leonie Wood, Hady Wardan, Jean-Francois Benoist, Lingli Li, Jim Vadolas, Joseph P. Sarsero, Panos A. Ioannou, Heidi L. Peters (Three first investigators contributed equally and should be considered as first authors)**; "**Gene induction for the treatment of Methylmalonic acidouria **"** ***The journal of gene Medicine*** , 2009, Volume 11, Issue 4, Page 361-369
3. DZ Loescha, **MS Khaniani** , HR Slaterd,e, JP Rubiof, QM Buig, K Kotscheth and at all Small CGG repeat expansion alleles of FMR1 gene are associated with Parkinsonism **Clinical Genetics** 2009: 76: 471–476
4. Danuta Z. Loesch, David E. Godler, **Mahmoud Khaniani**, Emma Gould and at all "Linking the FMR1 Alleles With Small CGG Expansions With Neurodevelopmental Disorders: Preliminary Data Suggest an Involvement of Epigenetic Mechanisms" **American Journal of Medical Genetics (Part A)**, 2009;10:2306-2310
5. A.G. Behbahan, B. Poorshiri, F. Mortazavi**, M.S. Khaniani** and S.M. Derakhshan "NPHS1 Gene Mutations in Children with Nephrotic Syndrome in Northwest Iran" **Pakistan Journal of Biological Sciences** 16 (17): 882-886, 2013
6. Ebrahimi Ammar, **khaniani S Mahmoud**, Mansoori Sima "Stable expression of modified Gene encoding functional Human coagulation Factor VIII" **Journal for Drugs and medicines,** 2011,Vol 3 (2), pp. 19-25
7. Negar Saliani, Masoud Darabi, Bahman Yousefi, Behzad Baradaran**, Mahmoud Shekari Khaniani**, Maryam Darabi, Maghsod Shaaker, Amir Mehdizadeh, Tahereh Naji and Mehrdad Hashemi. "PPARγ agonist-induced alterations in Δ6-desaturase and stearoyl-CoA desaturase 1: Role of MEK/ERK1/2 pathway" **World Journal of Hepatology,** 2013, 5(4): 220-225.

8. Bahman Yousefi, Masoud Darabi, Behzad Baradaran, **Mahmoud Shekari Khaniani**, Mohammad Rahbani, Maryam Darabi, Shabnam Fayezi, Amir Mehdizadeh, Negar Saliani, Maghsod Shaaker "Inhibition of MEK/ERK1/2 Signaling Affects the Fatty Acid Composition of HepG2 Human Hepatic Cell Line" **Bioimpacts, 2012, 2(3), 145-150**

9. -Jamal Mohmmadian. Sima Mansoori Derakhshan , Masood Mohmmadian, **Mahmoud Shekari Khaniani*** "Construction of Yeast Recombinant Expression Vector Containing Human Epidermal Growth Factor (hEGF)"
   Advanced Pharmaceutical Bulletin: 2013; 3(2), 473-476
10. Behrooz Farhadi, **Mahmoud Shekari Khaniani**, Sima Mansoori Derakhshan "Construction of pPIC9 Recombinant Vector Containing Human Stem Cell Factor" **Advanced Pharmaceutical Bulletin: 2013;3(2), 303-308**
11. **:** Azizeh Farshbaf khalili, Mahnaz Shahnazi, Khadijeh bHajizadeh, **Mahmoud Shekari Khaniani** "Down Syndrome Screening Methods In Iraniani Pregnant Women" **Journal of caing Scinces ,2012,(13),145-151**
12. Salman Asghari, **Mahmoud Shekari Khaniani** , Masood Darabei, Sima Mansoori Derakhshan "Cloning of Soluble Human Stem Cell Factor in pET-26b(+) vector"
    Advanced Pharmaceutical Bulletin, 2014, 4(1), 91-95
13. Sima Mansoori Derakhshan, Shamsei Abbasalizadeh, Fatemeh Abbasalizadeh, **Mahmoud Shekari Khaniani** * "Pernatal Diagnosis of Spinal Muscular Atrophy: Clinical experience and Moleculargenetics of SMN gene analysis in 36 cases"
    Journal of Prenatal Medicine, 2013 Vol: 7 (3); 32-34
14. **Mahmoud Shekari Khaniani**, Sima Mansoori Derakhshan **"**Molecular Characterisation and Assessment of Clinical Significance of Small Fragile X Alleles" **Journal of Analytical Research in Clinical Medicine (Vol. 1, No. 1 Page 1-17**)
15. Afkhamei F, **Shekari Khaniani .M**, Farzadei, Paknejade, Mansoori Derakhshan. S **"**The HLA- G 14bp insertion/delation polymorphisim in women with recurrent spontaneous abortion" **Iranian Journal of Allergy, Asthma and Immunology. 2014; 13 (5): 364-369**
16. Tamouchin Moharrami, Sima Mansoori Derakhshan, Abbas Ali H. Pourfeizi. **Mahmoud Shekari Khaniani "** Detaection of Haemaophillia A carriers in Azarei Turkish Population of Iran **Clinical and Applied Thrombosis /hemostasis (accepted- First online)** 2015,Vol2(8)755-759
17. Nahid Karimian fathi و **Mahmoud Shekari khaniani**, Vahid Motazeri , Sima Mansoori Derakhshan "Minor role of BRCA2 mutation (Exone 2 and Exone 11) in patients with early- onset breast cancer amongst Iranian Azari-Turkish women " **Iranian Journal of Basic Medical Sciences**, 2014 , Vol. 17 ,No 2

18. Samira Goldar, **Mahmoud Shekari Khaniani**, Sima Mansoori Derakhshan, Behzad Baradran " Molecular mechanisms Apoptosis and Role in cancer development and treatment" **Asian Pacific Journal of Cancer Prevention**: APJCP 2015, 16 (6): 2129-44

19. **Mahmoud Shekari Khaniani**, Nasrin Sohrapi, Neda Mansoori Derakhshan. Sima Mansoori Derakhshan " One novel frameshift mutation on exon 64 of COL7A1 gene in an Iranian individual suffering recessive Dystrophic epidermolysis Bullosa" **Annals of Clinical and Laboratory Sciences** Vol , 45 , no, 5 , 2015

20. Nasrin Sohrabi, **Mahmoud Shekari Khaniani**, Sima Mansoori Derakhshan " Evaluation of Association Between HLA Class II DR4–DQ8 Haplotype and Type I Diabetes Mellitus in Children of East Azerbaijan State of Iran" **Adv Pharm Bull**, 2015, Volume 17, Issue 2, February 2015, Page 108-111
21. Sima Mansoori Derakhshan1 , Aziz Khorrami2, Abbasali Hosseinpour Pour Feizi and **Mahmoud Shekari Khaniani** "Spectrum of β-Globin Gene Mutations and β-Thalassemia Haplotype Analysis among the Iranian Azeri Turkish Population " Epidemiology , 2015, Vol 5. Isuue 4
22. Sima Mansoori Derakhshan , Fatemeh Zeinali Sehrig, Nasrin Sohrabi, Siamak Shiva,4 Behzad Baradaran, and **Mahmoud Shekari Khaniani** " The Association between Human Leukocyte Antigen Class II DR3–DQ2 Haplotype and Type 1 Diabetes in Children of the East Azerbaijan State of Iran" **Iran Red Crescent Med J**. 2015 Sep; 17(9)
23. Neda Shahmohammadibeni • Simin Rahimi-Aliabadi • Javad Jamshidi3 abak Emamalizadeh • Hossein Ali Shahmohammadibeni • Alireza Zare Bidoki Haleh Akhavan-Niaki1 • Hajar Eftekhari1 • Shokoufeh Abdollahi6 **Mahmoud Shekari Khaniani** Mahnaz hahmohammadibeni•….. The analysis of association between SNCA, HUSEYO and CSMD1 gene variants and Parkinson's disease in Iranian population; **Neurological Sciences** (Volume 5- Issue 4) 2016

24. Mahmoud Shekari Khaniani, Mahdieh Taghizadeh, Abbasali Hosseinpour Feizi, Sima Mansoori Derakhshan" Sodium Butyrate and Valproic Acid as Splicing Restoring Agents in Erythroid Cells of b-Thalassemic Patients" **Iranian Journal of Biotechnology** , Volume 14, Issue 1, Winter 2016, Page 9-15

25. **Mahmoud Shekari Khaniani**, Sima Mansoori Derakhshan "Cytogenetic finding in patients with intellectual disability and/or multiple congenital anomalies" **Journal of Analytical Research in Clinical Medicine** (2016 ,Vol. 4, No. 2 Page 97-103)
26. **Mahmoud Shekari Khaniani**, Abdollah Ebrahimi, Setareh Daraei , Sima Mansoori Derakhshan "Genotyping of Intron Inversions and Point Mutations in Exon 14 of the FVIII Gene in Iranian Azeri Turkish Families with Hemophilia A" **Indian Journal of Hematology and Blood Transfusion**, December 2016, Volume 32, Issue 4, pp 475–480
27. **Mahmoud Shekar iKhaniani** , Fateme afkhamei, Fateme abbasalizadeh,, Sima Mansoori Derakhshan " Evaluation of thrombophilic Genes in Recurrent Pregnancy Loss: A Case- control Study in Iranian Women" International journal of Human Genetics, 2016 Volume 16(1.2): 48-52
28. S. Goldar1, B. Baradaran , **M. Shekari Khaniani**, A. Azadmehr, S. M. Derakhshan, A. Mohammadi, B. Mansoori, V. Khaze, D. Shanehbandi, L. Mohammadnejad, E. Baghbani " Extracts of Scrophularia frigida Boiss display potent antitumor effects in human breast cancer cells by inducing apoptosis and inhibition of expression of the human epidermal growth factor receptor 2" **Cellular and Molecular Biology** 2016, Volume 62 , Issue 9, 83-89
29. Ziba Garehdaghchi, Siam Mansoori Derakhshan, **Mahmoud Shekari Khaniani** " Evaluation of a newly discovered breast cancer susceptibility locus at 6q25.1 in Iranian Azari-Turkish women contemporary oncology 2016 ; 20(4): 308–310.
30. Faranak Madadi, **Mahmoud Shekari Khaniani**, Ehsan Esmaili Shandiz, Hormoz Ayromlou, Safa Najmi, Babak Emamalizadeh, Shaghayegh Taghavi, Javad Jamshidi, Abbas Tafakhori, Gholam-Ali Shahidi, Hossein Darvish5 " Genetic Analysis of the ZNF512B, SLC41A1, and ALDH2Polymorphisms in Parkinson's Diseasein the Iranian Population " **Genetic testing and Molecular Biomarkers** "2016 ;Volume 20(10):629-632
31. Soodabeh Khalili , **Mahmoud Shekari Khaniani** , Naser Aghamohammdzade, Abolfazl Akbarzadeh Sima Mansoori Derakhshan " The Association of Nucleobindin 2 Gene (NUCB2) Variants with Type 2 Diabetes Mellitus Among Iranian Azeri-Turkish Population" **Biosciences, Biotechnology Research Asia** 2016. Vol. 13(3), 1821-1828

32. Sima M. Derakhshan, **Mahmoud S. Khaniani**, Fateme Afkhami & Abbas Ali H. PourFeizi "Molecular Study of Deletional and Nondeletional Mutations on the α-Globin Locus in the Azeri Population of Northwestern Iran " **Hemoglbine**, 2016 VOL. 40, NO. 5, 319–322
33. Mohammad Reza Ranjouri, Parisa Aob, Sima Mansoori Derakhshan, **Mahmoud Shekari khaniani,** Hossein Chiti, Ali Ramazani " Association study of IL2RA and CTLA4 gene variants with type-1 diabetes mellitus in children in the northwest of Iran" **BioImpacts**, 2016, 6(4), 187-193
34. Sima Mansoori Derakhshan and **Mahmoud Shekari Khaniani** "Restoration of correct splicing in IVSI-110 mutation of β-globin gene with antisense oligonucleotides: implications and applications in functional assay development" Iranian Journal of Basic Medical Sciences. 2017, Vol 20, Issue 6, Page 700-707
35. Soodabeh Khalili, **Mahmoud Shekari Khaniani**, Fatemeh Afkhami, Sima Mansoori Derakhshan "NUCB2/Nesfatin-1: A Potent Meal Regulatory Hormone and its Role in Diabetes" **The Egyptian Journal of Medical Human Genetics** 2017,Volume 18, Issue 2, , Pages 105-109
36. , **Mahmoud Shekari Khaniani**, Parisa Abe,Mohmmadreza Ranjourei, Sima M. Derakhshan "Molecular analysis and prevalence of Huntington diseases in Northwestern Iran" **Turkish Journal of Medical Sciences**, Volume 47, 2017 -
37. Atefeh Entezari1• **Mahmoud Shekari Khaniani**, Tayyeb Bahrami, Sima Mansoori Derakhshan "Screening for intermediate CGG alleles of FMR1 gene in male Iranian patients with Parkinsonism" **Neurology Sciences** (2017) 38:123–128
38. **Shekari Khaniani Mahmoud** , Aziz Khoramei , Mandana Rafeey, Robabeh Gherghechi , Mansoori Derakhshan Sima Molecular Analysis of Glycogen Storage Disease type Ia in Iranian Azeri Turks: Identification of a Novel mutation" "**Journal of Genetics**" 2017, Mar;96(1):19-23
39. Tohid Ghasemnejad , **Mahmoud Shekari Khaniani** , , Fatemeh Zarei , Mina Farbodnia , Sima Mansoori Derakhshan " An update of common autosomal recessive non-syndromic hearing loss genes in Iranian population" **International Journal of Pediatric Otorhinolaryngology** 97 (2017) 113- 126
40. **Mahmoud Shekari Khaniani**, Fatemeh Amini Yeganeh , Shahrokh Amiri and Sima Mansouri Derakhshan, "Autistic Phenotype of Permutation and Intermediate Alleles of FMR1 Gene" Iranian Journal of Pediatric . 2017 ;27(4)):e9445. 2017 ;27(4)):e9445.

41. Sahar Bayat, **Mahmoud Shekari Khaniani**, Jalal Choupani, Mohammad Reza Alivand , Sima Mansoori Derakhshan, "HDACis (class I), cancer stem cell, and phytochemicals: Cancer therapy and prevention implications" **Biomedicine & Pharmacotherapy**, Volume 97, January, 2018, Pages 1445–1453
42. Jalal Choupani, Sima Mansouri Derakhshan, Sahar Bayat, Mohammad Reza Alivand and **Mahmood Shekari Khaniani**, "Narrower insight to SIRT1 role in cancer: A potential therapeutic target to control epithelial-mesenchymal transition in cancer cells" **Journal of cellular Physiology**, Volume 97, January 2018, Pages 1445–1453
43. Sepideh Zununi Vahed, Bahram Niknafs, **Mahmoud Shekari Khaniani**, Mohammadreza Ardalan " Genetic variations of complement factor H and C3 in patients with thrombotic thrombocytopenic purpura (TTP) in northwest of Iran" **Journal of Nephropathology** 2018 (in press)
44. Maryam Abtin, Mohammad R. Alivand, **Mahmoud S. Khaniani**, Milad Bastami Mohammad Zaeifizadeh, Sima M. Derakhshan," Simultaneous downregulation of miR-21 and miR-155 through oleuropein for breast cancer prevention and therapy **Journal of Cellular Biochemistryy** 2018(in press)
45. N. Yazdani, **M. Shekari Khaniani**, M. Bastami, T. Ghasemnejad, F. Afkhami, S. Mansoori Derakhshan," HLA-G regulatory variants and haplotypes with susceptibility to recurrent pregnancy loss " Internatinal of J Immunogenet. 2018;1–9.

Membership:

- Australasian Gene Therapy Society
- Human Genetics Society of Australia
- American Society of Human Genetics
- Human Genetics Society of Iran