

CURRICULUM VITAE

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Education:

- ❑ **May 2009- October 2011:** Postdoctoral fellowship- Department of Human Molecular Genetics, Max-Planck Institute for Molecular Genetics, Berlin, Germany.
- ❑ **January 2005- May 2009:** Ph.D. Department of Biology, Chemistry and Pharmacy, Free University, Berlin. Ph.D. thesis being completed in the Department of Human Molecular Genetics, Max-Planck Institute for Molecular Genetics, Berlin, Germany (magna Cum Laude).
- ❑ **September 2001- November 2003:** M.Sc. in human Genetics, Genetics Research Center, University of Welfare and Rehabilitation Sciences (USWR), Tehran, Iran, Grade: 18.54/20.
- ❑ **September 1997- July 2001:** B.Sc. in biology, Faculty of Sciences, Department of Biology, Ferdowsi University, Mashhad, Iran, Grade: 17.36/20.

Work Experiences

- ❑ **March 2019- present:** Associate professor, Department of Medical Genetics, faculty of Medical Sciences, Tarbiat Modares University, Tehran, Iran.
- ❑ **November 2011- March 2019:** assistant professor, Department of Medical Genetics, faculty of Medical Sciences, Tarbiat Modares University, Tehran, Iran.
- ❑ **January 2014-Present:** Establishment and head of Medical Genetics department at DeNA laboratory-Tehran Iran.
- ❑ **January 2012-Janury 2014:** Establishment and head of Medical Genetics department at Pars Hospital Lab-Tehran Iran.
- ❑ **July 2012-Present:** Establishment and head of Cytogenetics Lab at Masih Daneshvari Hospital- Shahid Beheshti University of Medical Sciences-Tehran-Iran.

Awards:

- ❑ First rank and honorary diploma from Academy of medical sciences Islamic republic of Iran in 2013.
- ❑ Ranked among the first 50 distinguished Iranian researchers in 2015 by the Iran Saramadane Elmi Federation (ISEF)
- ❑ Member of Iran's National Elites Foundation (INEF)
- ❑ Obtained fellowship from Max-Planck society for doing PhD from February 2004 to April 2008.
- ❑ Obtained fellowship from Max-Planck society for doing Postdoctoral research from May 2008 to October 2011.
- ❑ Obtained 10th position among all the Iranian students of biology which were attending in "8th OLYMPIAD of biology", May 2001.

Publications:

1: Imai-Okazaki A, Li Y, Horpaopan S, Riazalhosseini Y, **Garshasbi M**, Mosse YP, Zhang D, Schrauwen I, Sharma A, Fann CSJ, Leal SM, Lathrop M, Ott J. Heterozygosity mapping for human dominant trait variants. Hum Mutat. 2019 Apr 24. doi: 10.1002/humu.23765. [Epub ahead of print] PubMed PMID: 31018026.

- 2: Ji X, Li J, Huang Y, Sung PL, Yuan Y, Liu Q, Chen Y, Ju J, Zhou Y, Huang S, Chen F, Han Y, Yuan W, Fan C, Zhao Q, Wu H, Feng S, Liu W, Li Z, Chen J, Chen M, Yao H, Zeng L, Ma T, Fan S, Zhang J, Yuen KY, Cheng SH, Chik IWS, Liu NT, Zhu J, Lin S, Cao J, Tong S, Shan Z, Li W, Hekmat MR, **Garshasbi M**, Suela J, Torres Y, Cigudosa JC, Ruiz FJP, Rodríguez L, García M, Bernik J, Traven E, Reš U, Tul N, Tseng CF, Zhao D, Sun L, Pan Q, Shen L, Dai M, Wang Y, Wang J, Yang H, Yin Y, Duan T, Zhu B, Choolani M, Jin X, Chen Y, Mao M. Identifying occult maternal malignancies from 1.93 million pregnant women undergoing noninvasive prenatal screening tests. *Genet Med*. 2019 Apr 12. doi: 10.1038/s41436-019-0510-5. [Epub ahead of print] PubMed PMID: 30976098.
- 3: Karami J, Aslani S, Jamshidi A, **Garshasbi M**, Mahmoudi M. Genetic implications in the pathogenesis of rheumatoid arthritis; an updated review. *Gene*. 2019 Jun 20;702:8-16. doi: 10.1016/j.gene.2019.03.033. Epub 2019 Mar 20. Review. PubMed PMID: 30904715.
- 4: Kameli R, Ashrafi MR, Ehya F, Alizadeh H, Hosseinpour S, **Garshasbi M**, Tavasoli AR. Leukoencephalopathy in RIN2 syndrome: Novel mutation and expansion of clinical spectrum. *Eur J Med Genet*. 2019 Feb 13. pii: S1769-7212(18)30612-8. doi: 10.1016/j.ejmg.2019.02.002. [Epub ahead of print] PubMed PMID: 30769224.
- 5: Bitarafan F, **Garshasbi M**. Molecular Genetic Analysis of PKHD1 Mutations in Pedigrees With Autosomal Recessive Polycystic Kidney Disease. *Iran J Kidney Dis*. 2018 Nov;12(6):350-358. PubMed PMID: 30595564.
- 6: Hosseini Bereshneh A, **Garshasbi M**. Novel in-frame deletion in MFSD8 gene revealed by trio whole exome sequencing in an Iranian affected with neuronal ceroid lipofuscinosis type 7: a case report. *J Med Case Rep*. 2018 Sep 25;12(1):281. doi: 10.1186/s13256-018-1788-7. PubMed PMID: 30249282; PubMed Central PMCID: PMC6154911.
- 7: Mahmoudi M, **Garshasbi M**, Ashraf-Ganjouei A, Javinani A, Vojdani M, Saafi M, Ahmadzadeh N, Jamshidi A. Association between rs6759298 and Ankylosing Spondylitis in Iranian Population. *Avicenna J Med Biotechnol*. 2018 Jul-Sep;10(3):178-182. PubMed PMID: 30090213; PubMed Central PMCID: PMC6064007.
- 8: Razmara E, **Garshasbi M**. Whole-exome sequencing identifies R1279X of MYH6 gene to be associated with congenital heart disease. *BMC Cardiovasc Disord*. 2018 Jul 3;18(1):137. doi: 10.1186/s12872-018-0867-4. PubMed PMID: 29969989; PubMed Central PMCID: PMC6029398.
- 9: Daneshjoo O, **Garshasbi M**. Novel compound heterozygote mutations in the ATP7B gene in an Iranian family with Wilson disease: a case report. *J Med Case Rep*. 2018 Mar 15;12(1):68. doi: 10.1186/s13256-018-1608-0. PubMed PMID: 29540233; PubMed Central PMCID: PMC5853083.
- 10: Moteghaed NY, Maghooli K, **Garshasbi M**. Improving Classification of Cancer and Mining Biomarkers from Gene Expression Profiles Using Hybrid Optimization Algorithms and Fuzzy Support Vector Machine. *J Med Signals Sens*. 2018 Jan-Mar;8(1):1-11. PubMed PMID: 29535919; PubMed Central PMCID: PMC5840891.
- 11: Razmara E, Bitarafan F, Esmaeilzadeh-Gharehdaghi E, Almadani N, **Garshasbi M**. The first case of NSHL by direct impression on EYA1 gene and identification of one novel mutation in MYO7A in the Iranian families. *Iran J Basic Med Sci*. 2018 Mar;21(3):333-341. doi: 10.22038/IJBMS.2018.26269.6441. PubMed PMID: 29511501; PubMed Central PMCID: PMC5817178.
- 12: Bashti O, Noruzinia M, **Garshasbi M**, Abtahi M. miR-31 and miR-145 as Potential Non-Invasive Regulatory Biomarkers in Patients with Endometriosis. *Cell J*. 2018 Apr;20(1):84-89. doi: 10.22074/cellj.2018.4915. Epub 2017 Dec 1. Erratum in: *Cell J*. 2018 Jul;20(2):293. PubMed PMID: 29308623; PubMed Central PMCID: PMC5759684.
- 13: Ehya F, Abdul Tehrani H, **Garshasbi M**, Nabavi SM. Identification of miR-24 and miR-137 as novel candidate multiple sclerosis miRNA biomarkers using multi-staged data analysis protocol. *Mol Biol Res Commun*. 2017 Sep;6(3):127-140. doi: 10.22099/mbrc.2017.24861.1256. PubMed PMID: 29071282; PubMed Central PMCID: PMC5640895.
- 14: Inlora J, Sailani MR, Khodadadi H, Teymurinezhad A, Takahashi S, Bernstein JA, **Garshasbi M**, Snyder MP. Identification of a novel mutation in the APTX gene associated with ataxia-oculomotor apraxia. *Cold Spring Harb Mol Case Stud*. 2017 Nov 21;3(6). pii: a002014. doi: 10.1101/mcs.a002014. Print 2017 Nov. PubMed PMID: 28652255; PubMed Central PMCID: PMC5701303.
- 15: Hosseini Bereshneh A, Morshedi F, Hematyar M, Kaki A, **Garshasbi M**. Pharmacogenetics and Personalized Medicine in Pancreatic Cancer. *Acta Med Iran*. 2017 Mar;55(3):194-199. Review. PubMed PMID: 28282719.

- 16: Sailani MR, Jingga I, MirMazlomi SH, Bitarafan F, Bernstein JA, Snyder MP, **Garshasbi M**. Isolated Congenital Anosmia and CNGA2 Mutation. *Sci Rep*. 2017 Jun 1;7(1):2667. doi: 10.1038/s41598-017-02947-y. PubMed PMID: 28572688; PubMed Central PMCID: PMC5454015.
- 17: Musante L, Püttmann L, Kahrizi K, **Garshasbi M**, Hu H, Stehr H, Lipkowitz B, Otto S, Jensen LR, Tzschach A, Jamali P, Wienker T, Najmabadi H, Ropers HH, Kuss AW. Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. *Hum Mutat*. 2017 Jun;38(6):621-636. doi: 10.1002/humu.23205. Epub 2017 Mar 23. PubMed PMID: 28236339.
- 18: Khodadadi H, Azcona LJ, Aghamollai V, Omrani MD, **Garshasbi M**, Taghavi S, Tafakhori A, Shahidi GA, Jamshidi J, Darvish H, Paisán-Ruiz C. PTRHD1 (C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. *Mov Disord*. 2017 Feb;32(2):287-291. doi: 10.1002/mds.26824. Epub 2016 Oct 18. PubMed PMID: 27753167; PubMed Central PMCID: PMC5318269.
- 19: Aslani S, Mahmoudi M, **Garshasbi M**, Jamshidi AR, Karami J, Nicknam MH. Evaluation of DNMT1 gene expression profile and methylation of its promoter region in patients with ankylosing spondylitis. *Clin Rheumatol*. 2016 Nov;35(11):2723-2731. Epub 2016 Sep 16. PubMed PMID: 27637577.
- 20: Arabi E, **Garshasbi M**, Jamshidi AR, Khalesi R, Ahmadzadeh N, Akbarian M, Mahmoudi M. Association Study of an AFF1 Gene Polymorphism (rs340630) with Iranian Systemic Lupus Erythematosus Patients. *Acta Reumatol Port*. 2016 Jan-Mar;41(1):68-73. PubMed PMID: 27115110.
- 21: Momenzadeh P, Mahmoudi M, Beigy M, **Garshasbi M**, Vodjdanian M, Farazmand A, Jamshidi AR. Determination of IL1 R2, ANTXR2, CARD9, and SNAPC4 single nucleotide polymorphisms in Iranian patients with ankylosing spondylitis. *Rheumatol Int*. 2016 Mar;36(3):429-35. doi: 10.1007/s00296-015-3391-1. Epub 2015 Nov 21. PubMed PMID: 26590821.
- 22: Ataei-Kachouei M, Nadaf J, Akbari MT, Atri M, Majewski J, Riazalhosseini Y, **Garshasbi M**. Double Heterozygosity of BRCA2 and STK11 in Familial Breast Cancer Detected by Exome Sequencing. *Iran J Public Health*. 2015 Oct;44(10):1348-52. PubMed PMID: 26576347; PubMed Central PMCID: PMC4644579.
- 23: Heidari A, Tongsook C, Najafipour R, Musante L, Vasli N, **Garshasbi M**, Hu H, Mittal K, McNaughton AJ, Sritharan K, Hudson M, Stehr H, Talebi S, Moradi M, Darvish H, Arshad Rafiq M, Mozdhehipanah H, Rashidinejad A, Samiei S, Ghadami M, Windpassinger C, Gillissen-Kaesbach G, Tzschach A, Ahmed I, Mikhailov A, Stavropoulos DJ, Carter MT, Keshavarz S, Ayub M, Najmabadi H, Liu X, Ropers HH, Macheroux P, Vincent JB. Mutations in the histamine N-methyltransferase gene, HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. *Hum Mol Genet*. 2015 Oct 15;24(20):5697-710. doi: 10.1093/hmg/ddv286. Epub 2015 Jul 23. PubMed PMID: 26206890; PubMed Central PMCID: PMC4581600.
- 24: Iqbal Z, Püttmann L, Musante L, Razzaq A, Zahoor MY, Hu H, Wienker TF, **Garshasbi M**, Fattahi Z, Gilissen C, Vissers LE, de Brouwer AP, Veltman JA, Pfundt R, Najmabadi H, Ropers HH, Riazuddin S, Kahrizi K, van Bokhoven H. Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. *Eur J Hum Genet*. 2016 Mar;24(3):392-9. doi: 10.1038/ejhg.2015.148. Epub 2015 Jul 15. PubMed PMID: 26173967; PubMed Central PMCID: PMC4755381.
- 25: Moteghaed NY, Maghooli K, Pirhadi S, **Garshasbi M**. Biomarker Discovery Based on Hybrid Optimization Algorithm and Artificial Neural Networks on Microarray Data for Cancer Classification. *J Med Signals Sens*. 2015 Apr-Jun;5(2):88-96. PubMed PMID: 26120567; PubMed Central PMCID: PMC4460670.
- 26: Naghavi A, Mozdarani H, **Garshasbi M**, Yaghmaei M. Prevalence of Methylenetetrahydrofolate Reductase C677T Polymorphism in women with Polycystic Ovary Syndrome in southeast of Iran. *J Med Life*. 2015;8(Spec Iss 3):229-232. PubMed PMID: 28316696; PubMed Central PMCID: PMC5348926.
- 27: **Garshasbi M**, Ramazani A, Sorouri R, Javani S, Moradi S. Molecular detection of Brucella species in patients suspicious of Brucellosis from Zanjan, Iran. *Braz J Microbiol*. 2014 Aug 29;45(2):533-8. eCollection 2014. PubMed PMID: 25242938; PubMed Central PMCID: PMC4166279.
- 28: Larti F, Kahrizi K, Musante L, Hu H, Papari E, Fattahi Z, Bazazzadegan N, Liu Z, Banan M, **Garshasbi M**, Wienker TF, Ropers HH, Galjart N, Najmabadi H. A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. *Eur J Hum Genet*. 2015 Mar;23(3):331-6. doi: 10.1038/ejhg.2014.13. Epub 2014 Feb 26. Erratum in: *Eur J Hum Genet*. 2015 Mar;23(3):416. PubMed PMID: 24569606; PubMed Central PMCID: PMC4326716.

- 29: Püttmann L, Stehr H, **Garshasbi M**, Hu H, Kahrizi K, Lipkowitz B, Jamali P, Tzschach A, Najmabadi H, Ropers HH, Musante L, Kuss AW. A novel ALDH5A1 mutation is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. *Am J Med Genet A*. 2013 Aug;161A(8):1915-22. doi: 10.1002/ajmg.a.36030. Epub 2013 Jul 4. PubMed PMID: 23825041.
- 30: Papari E, Bastami M, Farhadi A, Abedini SS, Hosseini M, Bahman I, Mohseni M, **Garshasbi M**, Moheb LA, Behjati F, Kahrizi K, Ropers HH, Najmabadi H. Investigation of primary microcephaly in Bushehr province of Iran: novel STIL and ASPM mutations. *Clin Genet*. 2013 May;83(5):488-90. doi: 10.1111/j.1399-0004.2012.01949.x. Epub 2012 Sep 18. PubMed PMID: 22989186.
- 31: Kelly S, Pak C, **Garshasbi M**, Kuss A, Corbett AH, Moberg K. New kid on the ID block: neural functions of the Nab2/ZC3H14 class of Cys₃His tandem zinc-finger polyadenosine RNA binding proteins. *RNA Biol*. 2012 May;9(5):555-62. doi: 10.4161/rna.20187. Epub 2012 May 1. PubMed PMID: 22614829; PubMed Central PMCID: PMC3495735.
- 32: Abbasi-Moheb L, Mertel S, Gonsior M, Nouri-Vahid L, Kahrizi K, Cirak S, Wieczorek D, Motazacker MM, Esmaeeli-Nieh S, Cremer K, Weißmann R, Tzschach A, **Garshasbi M**, Abedini SS, Najmabadi H, Ropers HH, Sigrist SJ, Kuss AW. Mutations in NSUN2 cause autosomal-recessive intellectual disability. *Am J Hum Genet*. 2012 May 4;90(5):847-55. doi: 10.1016/j.ajhg.2012.03.021. Epub 2012 Apr 26. PubMed PMID: 22541559; PubMed Central PMCID: PMC3376487.
- 33: Najmabadi H, Hu H, **Garshasbi M**, Zemojtel T, Abedini SS, Chen W, Hosseini M, Behjati F, Haas S, Jamali P, Zecha A, Mohseni M, Püttmann L, Vahid LN, Jensen C, Moheb LA, Bienek M, Larti F, Mueller I, Weissmann R, Darvish H, Wrogemann K, Hadavi V, Lipkowitz B, Esmaeeli-Nieh S, Wieczorek D, Kariminejad R, Firouzabadi SG, Cohen M, Fattahi Z, Rost I, Mojahedi F, Hertzberg C, Dehghan A, Rajab A, Banavandi MJ, Hoffer J, Falah M, Musante L, Kalscheuer V, Ullmann R, Kuss AW, Tzschach A, Kahrizi K, Ropers HH. Deep sequencing reveals 50 novel genes for recessive cognitive disorders. *Nature*. 2011 Sep 21;478(7367):57-63. doi: 10.1038/nature10423. PubMed PMID: 21937992.
- 34: Hu H, Eggers K, Chen W, **Garshasbi M**, Motazacker MM, Wrogemann K, Kahrizi K, Tzschach A, Hosseini M, Bahman I, Hucho T, Mühlhoff M, Gerardy-Schahn R, Najmabadi H, Ropers HH, Kuss AW. ST3GAL3 mutations impair the development of higher cognitive functions. *Am J Hum Genet*. 2011 Sep 9;89(3):407-14. doi: 10.1016/j.ajhg.2011.08.008. PubMed PMID: 21907012; PubMed Central PMCID: PMC3169827.
- 35: Rafiq MA, Kuss AW, Püttmann L, Noor A, Ramiah A, Ali G, Hu H, Kerio NA, Xiang Y, **Garshasbi M**, Khan MA, Ishak GE, Weksberg R, Ullmann R, Tzschach A, Kahrizi K, Mahmood K, Naeem F, Ayub M, Moremen KW, Vincent JB, Ropers HH, Ansar M, Najmabadi H. Mutations in the alpha 1,2-mannosidase gene, MAN1B1, cause autosomal-recessive intellectual disability. *Am J Hum Genet*. 2011 Jul 15;89(1):176-82. doi: 10.1016/j.ajhg.2011.06.006. Erratum in: *Am J Hum Genet*. 2011 Aug 12;89(2):348. PubMed PMID: 21763484; PubMed Central PMCID: PMC3135808.
- 36: **Garshasbi M**, Kahrizi K, Hosseini M, Nouri Vahid L, Falah M, Hemmati S, Hu H, Tzschach A, Ropers HH, Najmabadi H, Kuss AW. A novel nonsense mutation in TUSC3 is responsible for non-syndromic autosomal recessive mental retardation in a consanguineous Iranian family. *Am J Med Genet A*. 2011 Aug;155A(8):1976-80. doi: 10.1002/ajmg.a.34077. Epub 2011 Jul 7. PubMed PMID: 21739581.
- 37: Pak C, **Garshasbi M**, Kahrizi K, Gross C, Apponi LH, Noto JJ, Kelly SM, Leung SW, Tzschach A, Behjati F, Abedini SS, Mohseni M, Jensen LR, Hu H, Huang B, Stahley SN, Liu G, Williams KR, Burdick S, Feng Y, Sanyal S, Bassell GJ, Ropers HH, Najmabadi H, Corbett AH, Moberg KH, Kuss AW. Mutation of the conserved polyadenosine RNA binding protein, ZC3H14/dNab2, impairs neural function in Drosophila and humans. *Proc Natl Acad Sci U S A*. 2011 Jul 26;108(30):12390-5. doi: 10.1073/pnas.1107103108. Epub 2011 Jul 6. PubMed PMID: 21734151; PubMed Central PMCID: PMC3145741.
- 38: Ropers F, Derivery E, Hu H, **Garshasbi M**, Karbasiyan M, Herold M, Nürnberg G, Ullmann R, Gautreau A, Sperling K, Varon R, Rajab A. Identification of a novel candidate gene for non-syndromic autosomal recessive intellectual disability: the WASH complex member SWIP. *Hum Mol Genet*. 2011 Jul 1;20(13):2585-90. doi: 10.1093/hmg/ddr158. Epub 2011 Apr 15. PubMed PMID: 21498477.
- 39: Timmermann B, Kerick M, Roehr C, Fischer A, Isau M, Boerno ST, Wunderlich A, Barmeyer C, Seemann P, Koenig J, Lappe M, Kuss AW, **Garshasbi M**, Bertram L, Trappe K, Werber M, Herrmann BG, Zatloukal K, Lehrach H, Schweiger MR. Somatic mutation profiles of MSI and MSS colorectal cancer identified by whole exome next generation sequencing and bioinformatics analysis. *PLoS One*. 2010 Dec 22;5(12):e15661. doi: 10.1371/journal.pone.0015661. PubMed PMID: 21203531; PubMed Central PMCID: PMC3008745.
- 40: Kuss AW, **Garshasbi M**, Kahrizi K, Tzschach A, Behjati F, Darvish H, Abbasi-Moheb L, Püttmann L, Zecha A, Weissmann R,

Hu H, Mohseni M, Abedini SS, Rajab A, Hertzberg C, Wieczorek D, Ullmann R, Ghasemi-Firouzabadi S, Banihashemi S, Arzhanghi S, Hadavi V, Bahrami-Monajemi G, Kasiri M, Falah M, Nikuei P, Dehghan A, Sobhani M, Jamali P, Ropers HH, Najmabadi H. Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. *Hum Genet.* 2011 Feb;129(2):141-8. doi: 10.1007/s00439-010-0907-3. Epub 2010 Nov 10. PubMed PMID: 21063731.

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42: Rivera-Brugués N, Albrecht B, Wieczorek D, Schmidt H, Keller T, Göhring I, Ekici AB, Tzschach A, **Garshasbi M**, Franke K, Klopp N, Wichmann HE, Meitinger T, Strom TM, Hempel M. Cohen syndrome diagnosis using whole genome arrays. *J Med Genet.* 2011 Feb;48(2):136-40. doi: 10.1136/jmg.2010.082206. Epub 2010 Oct 4. PubMed PMID: 20921020.

43: Kahrizi K, Hu CH, **Garshasbi M**, Abedini SS, Ghadami S, Kariminejad R, Ullmann R, Chen W, Ropers HH, Kuss AW, Najmabadi H, Tzschach A. Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. *Eur J Hum Genet.* 2011 Jan;19(1):115-7. doi: 10.1038/ejhg.2010.132. Epub 2010 Aug 11. PubMed PMID: 20700148; PubMed Central PMCID: PMC3039499.

44: Walczak-Sztulpa J, Eggenschwiler J, Osborn D, Brown DA, Emma F, Klingenberg C, Hennekam RC, Torre G, **Garshasbi M**, Tzschach A, Szczepanska M, Krawczynski M, Zachwieja J, Zwolinska D, Beales PL, Ropers HH, Latos-Bielenska A, Kuss AW. Cranioectodermal Dysplasia, Sensenbrenner syndrome, is a ciliopathy caused by mutations in the IFT122 gene. *Am J Hum Genet.* 2010 Jun 11;86(6):949-56. doi: 10.1016/j.ajhg.2010.04.012. Epub 2010 May 20. PubMed PMID: 20493458; PubMed Central PMCID: PMC3032067.

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48: Seifert W, Holder-Espinasse M, Kühnisch J, Kahrizi K, Tzschach A, **Garshasbi M**, Najmabadi H, Walter Kuss A, Kress W, Laureys G, Loeys B, Brilstra E, Mancini GM, Dollfus H, Dahan K, Apse K, Hennies HC, Horn D. Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants of COH1. *Hum Mutat.* 2009 Feb;30(2):E404-20. doi: 10.1002/humu.20886. PubMed PMID: 19006247.

49: Kahrizi K, Najmabadi H, Kariminejad R, Jamali P, Malekpour M, **Garshasbi M**, Ropers HH, Kuss AW, Tzschach A. An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. *Eur J Hum Genet.* 2009 Jan;17(1):125-8. doi: 10.1038/ejhg.2008.159. Epub 2008 Sep 10. PubMed PMID: 18781183; PubMed Central PMCID: PMC2985958.

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55: Dadgar S, Hagens O, Dadgar SR, Haghighi EN, Schimpf S, Wissinger B, **Garshasbi M**. Structural model of the OPA1 GTPase domain may explain the molecular consequences of a novel mutation in a family with autosomal dominant optic atrophy. *Exp Eye Res*. 2006 Sep;83(3):702-6. Epub 2006 May 12. PubMed PMID: 16698014.

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57: Khodayari N, **Garshasbi M**, Fadai F, Rahimi A, Hafizi L, Ebrahimi A, Najmabadi H, Ohadi M. Association of the dopamine transporter gene (DAT1) core promoter polymorphism -67T variant with schizophrenia. *Am J Med Genet B Neuropsychiatr Genet*. 2004 Aug 15;129B(1):10-2. PubMed PMID: 15274029.

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Technical Experiences:

- Establishment of Noninvasive prenatal testing (NIPT) using Ion Proton Next Generation Sequencing (NGS) at Medical Genetics department of DeNA Lab
- Establishment of following platforms for linkage analysis consisting of data conversion to linkage formats using two software packages (ALOHOMORA and easyLinkage), checking pedigree information [using gender check (ALOHOMORA) and Graphical Representation of Relationship errors (GRR)], detection of Mendelian and non-Mendelian Inheritance errors [using software PedCheck and Merlin], parametric single point linkage analysis (using software FastLink and SuperLink), parametric and non-parametric multipoint linkage analysis and haplotyping (using software Merlin, GeneHunter and Allegro) and visualization of haplotypes (using HaploPainter software).
- Familiar with molecular genetics techniques for instance: DNA extraction, RNA extraction, Agarose gel electrophoresis-PAGE/silver staining, SDS PAGE, PCR, RT PCR, ARMS (Amplification Refractory Mutation System) PCR, Reverse Dot blot, Rapid Amplification of cDNA ends (RACE), RNA interference (RNAi), STR Analysis, Restriction Fragment Length Polymorphism (RFLP), Southern blotting, Northern blotting, Western blotting, Cloning and Sub-cloning, cell culture, FIP-In-T-Rex, Chromatin Immunoprecipitation, Immunofluorescence and ...
- Familiar with Affymetrix SNP genotyping array, Illumina SNP genotyping array, Illumina expression profiling and relevant data analysis using Partek, BeadStudio and GTYPE.
- Familiar with cytogenetics techniques like: cell culture, chromosome analysis, Karyotyping, G banding, C-banding, Q-banding, HR banding, G11 banding and NOR banding.
- Familiar with different bioinformatics software and databases for prioritizing genes in linkage intervals, such as: Endeavour, Prioritizer, Posmed and ...

Published Books in Persian:

- ❑ Co-translator: Gunter Kahl. The Dictionary of Gene Technology. Wiley-VCH; second edition, published by Salemi publisher, Tehran, Iran (2005).
- ❑ Co-translator: Gardner A. Havel R. Davis T. Human genetics. Published by USWR publisher, Tehran, Iran (2005).
- ❑ Co- Author: Multiple-choice tests for genetics postgraduate entrance examinations, Published by Salemi publisher, Tehran, Iran, First edition (2002)
- ❑ Co- Author: Multiple-choice tests for genetics postgraduate entrance examinations, Published by Salemi publisher, Tehran, Iran, Second edition (2005).

Other activities:

- ❑ Contribution and presentation in the 16th International European congress of human genetics, Amsterdam RAI, The Netherlands, May 28 - 31, 2011
- ❑ Contribution and presentation in the 22 Annual Meeting German Human Genetics Society, 2011 in Regensburg, 16.-18.3.2011.
- ❑ Contribution and presentation in the 59th American Society of Human Genetics, 2009 Annual Meeting in Honolulu, Hawaii, USA, October 20-24.
- ❑ Contribution and presentation in the 21st Annual Meeting German Human Genetics Society, 2010 in Hamburg, Germany, March 2-4.
- ❑ Contribution in the second meeting of Iranians abroad, 10-12 Aug. 2010, Tehran, Iran.
- ❑ Contribution in the first meeting of Iranians abroad, 14-17 Apr 2009, Tehran, Iran.
- ❑ Contribution in the first biology conference for Iranian Scholars in Europe, 6-7 Dec 2008, Hamburg, Germany.
- ❑ Contribution in 13th International European congress of human genetics in conjunction with European Meeting on Psychosocial Aspects of Genetics (EMPAG), Barcelona, Spain. May 2008.
- ❑ Attendance in the 48th ANNUAL SHORT COURSE IN MEDICAL AND EXPERIMENTAL MAMMALIAN GENETICS, Joint program between Jackson Laboratory and John Hopkins University, Bar Harbor, Maine, USA. July 15-27, 2007.
- ❑ Contribution in 12th International European congress of human genetics, Nice, France. June 2007.
- ❑ Contribution in 10th International European congress of human genetics, Prague, Czech Republic. May 2005.
- ❑ Attendance in 18th Course in Medical Genetics held in Bertinoro di Romagna, Italy, May 15th - 21st, 2005
- ❑ Tutor for workshop "Large-scale elucidation of genetic diseases: data analysis and related issues" in the 18th Course in Medical Genetics, European school of genetics medicine, held in Bertinoro di Romagna, Italy, May 15th - 21st, 2005.
- ❑ Internal manager and editorial member for scientific newsletter of Genetic Research Center -USWR, (2001-2003).
- ❑ Contribution in genetics counseling course, organized by Nation-wide Genetics Counseling Network, Welfare Organization, Tehran, Iran (May 2003).
- ❑ Contribution and presentation in 8th Iranian congress of Genetics, 20-22 May 2003, Tehran, Iran.
- ❑ Participation in "Two days seminar on Human Genome Diversity in Islamic countries" held by National Institute for Genetic engineering and Biotechnology. 7-8 May 2002, Tehran, Iran.
- ❑ Collaboration in organizing and contribution in second Iranian Congress of Genetic Disorders and Disabilities (ICGDD), University of Social Welfare & Rehabilitation sciences, 7-11 Dec 2002, Tehran, IRAN.
- ❑ Participation in 8th congress of biology, Razi University, Kermanshah, Iran, June 1997.
- ❑ Contribution in the first congress of applied biology, Mashad, Iran, Aug. 2000.