University of Social Welfare and Rehabilitation Sciences

Ministry of Health and Medical Education

ITA		
Name: Hossein Surname: Najmabadi	Specialty /Ph.D. Molecular and Medical Genetics	
Title/Degree: Professor	Department of: Genetics/ Genetics Research Center	

Research Interests:

- Molecular Diagnostic of Single Gene Disorders
- Prenatal Diagnosis
- Gene Isolation, Cloning
- Transcriptional Regulation of Eukaryotic Genes

Scopus Profile: http://www.scopus.com/authid/detail.url?origin=resultslist&authorId=6701918454&zone= Google Scholar Profile: https://scholar.google.com/citations?user=YVvoh6IAAAAJ&hl=en	Updated: 2/24/2024

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Education

Date	Degree	Duration	Institution	Country/City	Major
1979-1983	BSc.	4 years	University of North Texas	Denton, Texas, USA	Biology
1980-1984	BSc.	4 years	University of North Texas	Denton, Texas, USA	Medical Technology
1984-1989	Ph.D.	5 years	University of North Texas	Denton, Texas, USA	Molecular Biology, Minors Human Genetics & Biochemistry
1990 –1995	Two Post Doc.	5 years	UCLA Medical Center	Torrance, CA, USA	Transcriptional Regulation of Inhibin Gene, and Mapping of Yq Microdeletion in oligozoospermic men

Faculty member

Year	Position	Duration	Institution/Course	Location
2006 - present	Professor, Head & Director of GRC	15 years	Genetics Research Center (GRC), University of Social Welfare & Rehabilitation Sciences (USWR)	Tehran, Iran
2002 - 2006	Associate Professor, Head & Director of GRC	4 years	GRC, USWR	Tehran, Iran
1996 - 2002	Assistant Professor, Head & Director of GRC	6 years	GRC, USWR	Tehran, Iran
1995 - 1996	Assistant Professor of Medicine	1 year	Charles Drew University of Medicine & Science – UCLA, Los Angeles	CA, USA
1990 - 1995	Postdoctoral Fellow	5 years	Charité – Universitätsmedizin Berlin / INFOGEN	CA, USA

Field of Specialization

Molecular Diagnostic of Single Gene Disorders

Prenatal Diagnosis

Gene Isolation, Cloning

Transcriptional Regulation of Eukaryotic Genes

Language Ability

- English

Research Experience

Year	Position	Institution/Course	Location
2017- present	Establishment & Director, Iranian Population Database "Iranome (www.iranome.com)	University of Social Welfare and Rehabilitation Sciences	Tehran, Iran
2003-Present	Establishment & Director, Iranian Human Gene Bank	University of Social Welfare and Rehabilitation Sciences	Tehran, Iran
1996 - Present	Director of Molecular Division	Kariminejad - Najmabadi Phatology & Genetics Center	Tehran, Iran

Scientific Membership

Year	**Association, Society**	**Location**
2011 – present	Editorial Board, Archive of Iranian Medicine	Tehran, Iran
2011- present	Editorial Board, Clinical Genetics	USA
2011-Present	Educational Board (PhD Degree), University of Social Welfare and Rehabilitation Sciences	Tehran, Iran
2010 -present	Member, Association for Molecular Pathology (AMP)	USA
2006-Present	Member, European Society of Human Genetics (ESHG)	Vienna, Austria
1996 - present	Member, American Society of Human Genetics (ASHG)	USA
1980 - present	Member, American Society of Clinical Pathology (ASCP)	USA

Selected Publications:

1. Abolhassani A, Fattahi Z, Beheshtian M, Fadaee M, Vazehan R, Ahangari F, Dehdahsi S, Faraji Zonooz M, Parsimehr E, Kalhor Z, Peymani F. Clinical application of next generation sequencing for Mendelian disease diagnosis in the Iranian population. *npj Genomic Medicine*. 2024 Feb 19;9(1):12.

2. Mehvari S, Karimian Fathi N, Saki S, Asadnezhad M, Arzhangi S, Ghodratpour F, Mohseni M, Zare Ashrafi F, Sadeghian S, Boroumand M, Shokohizadeh F. Contribution of genetic variants in the development of familial premature coronary artery disease in a cohort of cardiac patients. *Clinical Genetics*. 2024 Feb 3.

3. Rashvand Z, **Najmabadi H**, Kahrizi K, Mozhdehipanah H, Moradi M, Estaki Z, Taherkhani K, Nikzat N, Najafipour R, Omrani MD. Identification of a Novel Variant in CC2D1A Gene Linked to Autosomal Recessive Intellectual Disability 3 in an Iranian Family and Investigating the Structure and Pleiotropic Effects of this Gene. Iranian Journal of *Child Neurology*. 2024 Jan 18;18(1):25-41.

4. Bazazzadegan, N., Abedini, S. S., Azarkeivan, A., Banihashemi, S., Nikzat, N., **Najmabadi, H**., & Neishabury, M. (2023). The Spectrum of HBB Mutations among 2315 Beta Thalassemia Patients of a Reference Clinic in Tehran-Iran.

Hemoglobin, 1-5.

5. Pagnamenta, A.T., Belles, R.S., Salbert, B.A., Wentzensen, I.M., Guillen Sacoto, M.J., Santos, F.J.R., Caffo, A., Ferla, M., Banos-Pinero, B., Pawliczak, K. and Makvand, M., 2023. The prevalence and phenotypic range associated with biallelic PKDCC variants. *Clinical Genetics*.

6. Moghadam, M. G., Elahi, Z., Soveyzi, M., Arzhangi, S., Nafissi, S., **Najmabadi, H**., ... & Fattahi, Z. (2023). Expanding the Molecular Spectrum of HK1-Related Charcot-Marie-Tooth Disease, Type 4G; the First Report in Iran. *Archives of Iranian Medicine (AIM)*, *26*(5).

7. Jamshidi, F., Shokouhian, E., Mohseni, M., Kahrizi, K., **Najmabadi, H**., & Babanejad, M. (2023). Identification of a homozygous frameshift mutation in the FGF3 gene in a consanguineous Iranian family: First report of labyrinthine aplasia, microtia, and microdontia syndrome in Iran and literature review. *Molecular Genetics & Genomic Medicine*, *11*(5), e2168.

8. Ashrafi, F. Z., Akhtarkhavari, T., Fattahi, Z., Asadnezhad, M., Beheshtian, M., Arzhangi, S., ... & Kahrizi, K. (2023). Emerging Epidemiological Data on Rare Intellectual Disability Syndromes from Analyzing the Data of a Large Iranian Cohort. *Archives of Iranian Medicine*, *26*(4), 186-197.

9. Elahi, Z., Soveyzi, M., Nafissi, S., Nilipour, Y., Goleyjani Moghadam, M., Keshavarz, E., Kariminejad, A., **Najmabadi, H**. and Fattahi, Z., 2023. Bi-allelic loss of function variant in the NRCAM gene is associated with motor-predominant axonal polyneuropathy; the second report. *Molecular Genetics & Genomic Medicine*, p.e2131.

10. Mohseni, M., Mohammadi, Y., Ashrafi, F. Z., Ghodratpour, F., Jalalvand, K., Arzhangi, S., ... & **Najmabadi, H**. (2023). An Extended Iranian Family with Autosomal Dominant Non-syndromic Hearing Loss Associated with A Nonsense Mutation in the DIAPH1 Gene. *Archives of Iranian Medicine*, *26*(3), 176-180.

11. Dawood, M., Akay, G., Mitani, T., Marafi, D., Fatih, J.M., Gezdirici, A., **Najmabadi, H**., Kahrizi, K., Punetha, J., Grochowski, C.M. and Du, H., 2023. A biallelic frameshift indel in PPP1R35 as a cause of primary microcephaly. *American Journal of Medical Genetics Part A*.

12. Mensah, M.A., Niskanen, H., Magalhaes, A.P., Basu, S., Kircher, M., Sczakiel, H.L., Reiter, A.M., Elsner, J., Meinecke, P., Biskup, S. and Chung, B.H., 2023. Aberrant phase separation and nucleolar dysfunction in rare genetic diseases. *Nature*, pp.1-8.

13. Ashrafi, F. Z., Mohseni, M., Beheshtian, M., Fattahi, Z., Ghodratpour, F., Keshavarzi, F., ... & **Najmabadi, H**. (2023). Implementation of an In-House Platform for Rapid Screening of SARS-CoV-2 Genome Variations. *Archives of Iranian Medicine*, *26*(2), 69-75.

14. Mahmoudi-Aznaveh A, Tavoosidana G, **Najmabadi H**, Azizi Z, Ardestani A. The liver-derived exosomes stimulate insulin gene expression in pancreatic beta cells under condition of insulin resistance. *Frontiers in Endocrinology*. 2023; 14.

15. Ehtesham, N., Mosallaei, M., Beheshtian, M., Khoshbakht, S., Fadaee, M., Vazehan, R., ... & **Najmabadi, H**. (2022). Characterizing Genotypes and Phenotypes Associated with Dysfunction of Channel-Encoding Genes in a Cohort of Patients with Intellectual Disability. *Archives of Iranian Medicine*, *25*(12), 788-797.

16. Saghi, M., InanlooRahatloo, K., Alavi, A., Kahrizi, K., **Najmabadi, H**. Intellectual disability associated with craniofacial dysmorphism due to POLR3B mutation and defect in spliceosomal machinery.BMC Medical Genomics, 2022,15(1),89.

17. Sisakht, J. M., Mehri, M., **Najmabadi, H**., Azarkeivan, A., & Neishabury, M. (2022). Genetic Diagnosis of Pyruvate Kinase Deficiency in Undiagnosed Iranian Patients with Severe Hemolytic Anemia, using Whole Exome Sequencing. *Archives of Iranian Medicine*, *25*(10), 691.

18. Hosseinpour, M., Ardalani, F., Mohseni, M., Beheshtian, M., Arzhangi, S., Ossareh, S., ... & Broumand, B. (2022). Targeted Next Generation Sequencing Revealed Novel Variants in the PKD1 and PKD2 Genes of Iranian Patients with Autosomal Dominant Polycystic Kidney Disease. *Archives of Iranian Medicine*, *25*(9), 600-608.

19. Hashemi, S. S., Hajati, R., Davarzani, A., Rohani, M., DanaeeFard, F., Bidgoli, M. M. R., ... & Alavi, A. (2022). Anticipation can be more common in hereditary spastic paraplegia with SPAST mutations than it appears. *Canadian Journal of Neurological Sciences*, *49*(5), 651-661.

20. Karimzadeh, P., **Najmabadi, H**., Lochmuller, H., Babaee, M., Dehdahsi, S., Miryounesi, M., ... & Tonekaboni, S. H. (2022). Five patients with spinal muscular atrophy-progressive myoclonic epilepsy (SMA-PME): a novel pathogenic variant, treatment and review of the literature. *Neuromuscular Disorders*.

21. Goodarzi, A., Rad, F., Esmaeili, O. S., Forouzeshpour, F., **Najmabadi, H**., & Azarkeivan, A. (2022). 3091– CLINICAL FEATURES AND MOLECULAR BASIS OF THALASSEMIC PATIENTS WITH HOMOZYGOTE FORM OF IVSII-I/IVSII-I MUTATION AND SYNCHRONIZATION WITH A-GENE MUTATION. *Experimental Hematology*, *111*, S90.

22. Dulika Sumathipala, Petter Strømme , Zohreh Fattahi , Torben Lüders, Ying Sheng , Kimia Kahrizi , Ingunn Holm Einarsen, Jennifer L Sloan , **Hossein Najmabadi** , Lambert van den Heuvel, Ron A Wevers, Sergio Guerrero-Castillo, Lars Mørkrid, Vassili Valayannopoulos, Paul Hoff Backe, Charles P Venditti , Clara D van Karnebeek , Hilde Nilsen , Eirik Frengen, Doriana Misceo. ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences.Brain, 2022 Jul 29;145(7):2602-2616

23. Fattahi, Z., Mohseni, M., Jalalvand, K., (...), Jazayeri, S.M., **Najmabadi, H**, SARS-CoV-2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. Transboundary and Emerging Diseases, 2022, 69(3), pp

24. Fattahi Z, Mohseni M, Beheshtian M, (…), Jazayeri SM, **Najmabadi H.** Disease Waves of SARS-CoV-2 in Iran Closely Mirror Global Pandemic Trends. Arch Iran Med. 2022 Aug 1;25(8):508-522.

25. Sadighi, G., Astaneh, A.N., **Najmabadi, H.**, Ardakani, M.R.K., Latifi-Navid, S. DNA Banking to Assess Genetic Influences on Schizophrenia. Medical Journal of the Islamic Republic of Iran, 2022, 36(1).

26. Rashvand, Z., Kahrizi, K., **Najmabadi, H**., Najafipour, R., Omrani, M.D. A novel variant of C12orf4 linked to autosomal recessive intellectual disability type 66 with phenotype expansion. Journal of Gene Medicine, 2022, 24(4),e3406.

27. Mosallaei, M., Ehtesham, N., Beheshtian, M., (...), Kahrizi, K., **Najmabadi, H**. Phenotype and genotype spectrum of variants in guanine nucleotide exchange factor genes in a broad cohort of Iranian patients. Molecular Genetics and Genomic Medicine, 2022, 10(4),e1894.

28. Sotoudeh Anvari, M., Vasei, H., **Najmabadi, H**., (...), Goodarzynejad, H., Mowla, S.J. Identification of microRNAs associated with human fragile X syndrome using next-generation sequencing. Scientific Reports, 2022, 12(1),5011.

29. Hashemieh, M., Saadatmandi, Z.A.S., Azarkeivan, A., **Najmabadi, H.** The Effect of Xmn-1 Polymorphism and Coinheritance of Alpha Mutations on Age at First Blood Transfusion in Iranian Patients with Homozygote IVSI-5 Mutation, International Journal of Hematology-Oncology and Stem Cell Research, 2022, 16(1), pp. 47-54

30. Babanejad, M., Beheshtian, M., Jamshidi, F., (...), Kahrizi, K., **Najmabadi, H**. Genetic etiology of hearing loss in Iran. Human Genetics, 2022, 141(3-4), pp. 623-631

31. Fazeli, Z., Ghaderian, S.M.H., **Najmabadi, H**., Omrani, M.D. Understanding the Molecular Basis of Fragile X Syndrome Using Differentiated Mesenchymal Stem Cells.Iranian Journal of Child Neurology, 2022, 16(1), pp. 85-95

32. Kariminejad A, Shakiba M, Shams M, Namiranian P, Eghbali M, Talebi S, Makvand M, Jaeken J, **Najmabadi H**,

Hennekam RC. NGLY1 deficiency: Novel variants and literature review. Eur J Med Genet. 2021; 64(3):104146.

33. Neishabury M, Azarkeivan A, Mehri M, **Najmabadi H**, Cheraghi T. The first case of BENTA disease (B cell expansion with NF-κB and T cell anergy) from Iran. J Clin Immunol. 2021; 41(4):811-3.

34. Oláhová M, Peter B, Szilagyi Z, Diaz-Maldonado H, Singh M, Sommerville EW, Blakely EL, Collier JJ, Hoberg E, Stránecký V, Hartmannová H, Bleyer AJ, McBride KL, Bowden SA, Korandová Z, Pecinová A, Ropers HH, Kahrizi K, **Najmabadi H**, Tarnopolsky MA, Brady LI, Weaver KN, Prada CE, Õunap K, Wojcik MH, Pajusalu S, Syeda SB, Pais L, Estrella EA, Bruels CC, Kunkel LM, Kang PB, Bonnen PE, Mráček T, Kmoch S, Gorman GS, Falkenberg M, Gustafsson CM, Taylor RW. POLRMT mutations impair mitochondrial transcription causing neurological disease. Nat Commun. 2021; 12(1):1135.

35. Beheshtian M, Akhtarkhavari T, Mehvari S, Mohseni M, Fattahi Z, Abedini SS, Arzhangi S, Fadaee M, Jamali P, Najafipour R, Kalscheuer VM, Hu H, Ropers HH, **Najmabadi H**, Kahrizi K. Comprehensive genotype-phenotype correlation in AP-4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clin Genet. 2021; 99(1):187-92.

36. Rahimi Bidgoli MM, Javanparast L, Rohani M, **Najmabadi H**, Zamani B, Alavi A. CAPN1 and hereditary spastic paraplegia: a novel variant in an Iranian family and overview of the genotype-phenotype correlation. Int J Neurosci. 2021; 131(10):962-74.

37. Najafi K, Mehrjoo Z, Ardalani F, Ghaderi-Sohi S, Kariminejad A, Kariminejad R, **Najmabadi H**. Identifying the causes of recurrent pregnancy loss in consanguineous couples using whole exome sequencing on the products of miscarriage with no chromosomal abnormalities. Sci Rep. 2021; 11(1):6952.

38. Jonkhout N, Cruciani S, Santos Vieira HG, Tran J, Liu H, Liu G, Pickford R, Kaczorowski D, Franco GR, Vauti F, Camacho N, Abedini SS, **Najmabadi H**, Ribas de Pouplana L, Christ D, Schonrock N, Mattick JS, Novoa EM. Subcellular relocalization and nuclear redistribution of the RNA methyltransferases TRMT1 and TRMT1L upon neuronal activation. RNA Biology, 2022, 18(11), pp. 1905-1919

39. Mohseni M, Babanejad M, Booth KT, Jamali P, Jalalvand K, Davarnia B, Ardalani F, Khoshaeen A, Arzhangi S, Ghodratpour F, Beheshtian M, Jahanshad F, Otukesh H, Bahrami F, Seifati SM, Bazazzadegan N, Habibi F, Behravan H, Mirzaei S, Keshavarzi F, Nikzat N, Mehrjoo Z, Thiele H, Nothnagel M, Azaiez H, Smith RJ, Kahrizi K, **Najmabadi H**. Exome sequencing utility in defining the genetic landscape of hearing loss and novel-gene discovery in Iran. Clin Genet. 2021; 100(1):59-78.

40. Diaz, J., Gerard, X., Emerit, M. B., Areias, J., Geny, D., Degardin, J., ... & Masson, J. (2021). YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations (vol 143, pg 2911, 2020). *BRAIN*, *144*.

41. Nair, D., Li, D., Erdogan, H., (...) **Najmabadi, H**, Ropers, H.-H., Bhoj, E.J. Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances,2021, 2(2),100024.

42. Averdunk, L., Sticht, H., Surowy, H., (...), **Najmabadi, H**., Alkuraya, F.S., Wieczorek, D. Correction to: The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype (Journal of Molecular Medicine, (2021), 99, 12, (1755-1768), 10.1007/s00109-021-02124-9). Journal of Molecular Medicine, 2021,99(12), pp. 1769-1770

43. Cheraghi, S., Moghbelinejad, S., **Najmabadi, H**., Kahrizi, K., Najafipour, R. The PTRHD1 mutation in intellectual disability. Archives of Iranian Medicine, 2021, 24(10), pp. 747-751

44. Averdunk L, Sticht H, Surowy H, Lüdecke HJ, Koch-Hogrebe M, Alsaif HS, Kahrizi K, Alzaidan H, Alawam BS, Tohary M, Kraus C, Endele S, Wadman E, Kaplan JD, Efthymiou S, **Najmabadi H**, Reis A, Alkuraya FS, Wieczorek D. The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype.

Journal of Molecular Medicine,2021.

45. Sheikh TI, Vasli N, Pastore S, Kharizi K, Harripaul R, Fattahi Z, Pande S, Naeem F, Hussain A, Mir A, Islam O, Girisha KM, Irfan M, Ayub M, Schwarzer C, **Najmabadi H**, Shukla A, Sladky VC, Braun VZ, Garcia-Carpio I, Villunger A, Vincent JB. Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. Transl Psychiatry. 2021; 11(1):1.

46. Rashvand Z, Kahrizi K, **Najmabadi H**, Najafipour R, Omrani MD. Clinical and genetic characteristics of splicing variant in CYP27A1 in an Iranian family with cerebrotendinous xanthomatosis. Iran Biomed J. 2021; 25(2):132-9.

47. Atroshi SD, Al-Allawi N, Chui DHK, **Najmabadi H**, Khailany RA. A novel β0-thalassemia mutation, HBB: c.356_357delTT [Codon 118 (-TT)] in an Iraqi Kurd. Hemoglobin. 2021; 45(3):212-4.

48. Khoshbakht S, Beheshtian M, Fattahi Z, Bazazzadegan N, Parsimehr E, Fadaee M, Vazehan R, Faraji Zonooz M, Abolhassani A, Makvand M, Kariminejad A, Celik A, Kahrizi K, **Najmabadi H**. CEP104 and CEP290; genes with ciliary functions cause intellectual disability in multiple families. Arch Iran Med. 2021; 24(5):364-73.

49. Mehrjoo Z, Kahrizi K, Mohseni M, Akbari M, Arzhangi S, Jalalvand K, **Najmabadi H**, Farhadi M, Mohseni M, Asghari A, Mohebbi S, Daneshi A. Limbic system associated membrane protein mutation in an Iranian family diagnosed with Ménière's disease. Arch Iran Med. 2020; 23(5):319-325.

50. Kazemi G, Peymani F, Mohseni M, Zare Ashrafi F, Arzhangi S, Ardalani F, Aghakhani Moghaddam F, Kahrizi K, **Najmabadi H**. Novel Mutation in LARP7 in Two Iranian Consanguineous Families with Syndromic Intellectual Disability and Facial Dysmorphism. Arch Iran Med. 2020; 23(12):842-7.

51. Forouzesh Pour F, Karimi K, Ghaderi Z, Tavakoli Koudehi A, **Najmabadi H**. Heterozygosity for the novel HBA2: c.*91_*92delTA polyadenylation site variant on the α2-globin gene expanding the genetic spectrum of α-thalassemia in Iran. Hemoglobin. 2020; 44(6):423-6.

52. Diaz J, Gérard X, Emerit MB, Areias J, Geny D, Dégardin J, Simonutti M, Guerquin MJ, Collin T, Viollet C, Billard JM, Métin C, Hubert L, Larti F, Kahrizi K, Jobling R, Agolini E, Shaheen R, Zigler A, Rouiller-Fabre V, Rozet JM, Picaud S, Novelli A, Alameer S, **Najmabadi H**, Cohn R, Munnich A, Barth M, Lugli L, Alkuraya FS, Blaser S, Gashlan M, Besmond C, Darmon M, Masson J. YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain. 2020; 143(10):2911-28.

53. Safari S, Ebadifar A, **Najmabadi H**, Kamali K, Abedini SS. Screening for PAX9, MSX1 and WNT10A mtations in 4 Iranian families with non-syndromic tooth agenesis. Avicenna J Med Biotechnol. 2020; 12(4):236-40.

54. Neishabury M, Mehri M, Fattahi Z, **Najmabadi H**, Azarkeivan A. Novel variants in Iranian individuals suspected to have inherited red blood cell disorders, including bone marrow failure syndromes. Haematologica. 2020; 105(1):e1-e4.

55. Taghizadeh S, Vazehan R, Beheshtian M, Sadeghinia F, Fattahi Z, Mohseni M, Arzhangi S, Nafissi S, Kariminejad A, **Najmabadi H**, Kahrizi K. Molecular diagnosis of hereditary neuropathies by whole exome sequencing and expanding the phenotype spectrum. Arch Iran Med. 2020; 23(7):426-33.

56. Fatehi F, Okhovat AA, Nilipour Y, Mroczek M, Straub V, Töpf A, Palibrk A, Peric S, Rakocevic Stojanovic V, **Najmabadi H**, Nafissi S. Adult-onset very-long-chain acyl-CoA dehydrogenase deficiency (VLCADD). Eur J Neurol. 2020; 27(11):2257-66.

57. Mehvari S, Larti F, Hu H, Fattahi Z, Beheshtian M, Abedini SS, Arzhangi S, Ropers HH, Kalscheuer VM, Auld D, Kahrizi K, Riazalhosseini Y, **Najmabadi H**. Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Mol Genet Genomic Med. 2020; 26:e1418.

58. Bowers M, Liang T, Gonzalez-Bohorquez D, Zocher S, Jaeger BN, Kovacs WJ, Röhrl C, Cramb KML, Winterer J,

Kruse M, Dimitrieva S, Overall RW, Wegleiter T, **Najmabadi H**, Semenkovich CF, Kempermann G, Földy C, Jessberger S. FASN-dependent lipid metabolism links neurogenic stem/progenitor cell activity to learning and memory deficits. Cell Stem Cell. 2020; 27(1):98-109.e11.

59. Mohseni M, Akbari M, Booth KT, Babanejad M, Azaiez H, Ardalani F, Arzhangi S, Jalalvand K, Nikzat N, Ghodratpour F, Jamali P, Adeli OA, Habibi H, Kahrizi K, **Najmabadi H**. When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). J Hum Genet. 2020; 65(7):609-17.

60. Rostami P, Zendehdel K, Shirkoohi R, Ebrahimi E, Ataei M, Imanian H, **Najmabadi H**, Akbari MR, Sanati MH. Gene panel testing in hereditary breast cancer. Arch Iran Med. 2020; 23(3):155-62.

61. Kariminejad A, Ghaderi-Sohi S, Keshavarz E, Hashemi SA, Parsimehr E, Szenker-Ravi E, Khatoo M, Faraji Zonooz M, Reversade B, **Najmabadi H**, Hennekam RC. A GLI3 variant leading to polydactyly in heterozygotes and Pallister-Hall-like syndrome in a homozygote. Clin Genet. 2020; 97(6):915-9.

62. Pashaei M, Rahimi Bidgoli MM, Zare-Abdollahi D, **Najmabadi H**, Haji-Seyed-Javadi R, Fatehi F, Alavi A. The second mutation of SYCE1 gene associated with autosomal recessive nonobstructive azoospermia. J Assist Reprod Genet. 2020; 37(2):451-8.

63. Cheraghi S, Moghbelinejad S, **Najmabadi H**, Kahrizi K, Najafipour R. A novel PTC mutation in the BTB domain of KLHL7 gene in two patients with Bohring-Opitz syndrome-like features. Eur J Med Genet. 2020; 63(4):103849.

64. Fadaee M, Mohseni M, Zeinali F, Shokouhi J, Fatahi Z, **Najmabadi H**, Kahrizi K. The second Iranian family with a known missense mutation in AP4M1, precise clinical characterization conducted to AP4 deficiency syndrome. (Submitted, Iranian Red Crescent Medical Journal)

65. Hu H, Kahrizi K, Musante L, Fattahi Z, Herwig R, Hosseini M, Oppitz C, Abedini SS, Suckow V, Larti F, Beheshtian M, Lipkowitz B, Akhtarkhavari T, Mehvari S, Otto S, Mohseni M, Arzhangi S, Jamali P, Mojahedi F, Taghdiri M, Papari E, Soltani Banavandi MJ, Akbari S, Tonekaboni SH, Dehghani H, Ebrahimpour MR, Bader I, Davarnia B, Cohen M, Khodaei H, Albrecht B, Azimi S, Zirn B, Bastami M, Wieczorek D, Bahrami G, Keleman K, Vahid LN, Tzschach A, Gärtner J, Gillessen-Kaesbach G, Varaghchi JR, Timmermann B, Pourfatemi F, Jankhah A, Chen W, Nikuei P, Kalscheuer VM, Oladnabi M, Wienker TF, Ropers HH, **Najmabadi H**. Genetics of intellectual disability in consanguineous families. Mol Psychiatry. 2019; 24(7):1027-39.

66. Kahrizi K, Hu H, Hosseini M, Kalscheuer VM, Fattahi Z, Beheshtian M, Suckow V, Mohseni M, Lipkowitz B, Mehvari S, Mehrjoo Z, Akhtarkhavari T, Ghaderi Z, Rahimi M, Arzhangi S, Jamali P, Falahat Chian M, Nikuei P, Sabbagh Kermani F, Sadeghinia F, Jazayeri R, Tonekaboni SH, Khoshaeen A, Habibi H, Pourfatemi F, Mojahedi F, Khodaie-Ardakani MR, Najafipour R, Wienker TF, **Najmabadi H**, Ropers HH. Effect of inbreeding on intellectual disability revisited by trio sequencing. Clin Genet. 2019; 95(1):151-9.

67. Mehrjoo Z, Fattahi Z, Beheshtian M, Mohseni M, Poustchi H, Ardalani F, Jalalvand K, Arzhangi S, Mohammadi Z, Khoshbakht S, Najafi F, Nikuei P, Haddadi M, Zohrehvand E, Oladnabi M, Mohammadzadeh A, Jafari MH, Akhtarkhavari T, Gooshki ES, Haghdoost A, Najafipour R, Niestroj LM, Helwing B, Gossmann Y, Toliat MR, Malekzadeh R, Nürnberg P, Kahrizi K, **Najmabadi H**, Nothnagel M. Distinct genetic variation and heterogeneity of the Iranian population. PloS Genet. 2019; 15(9):e1008385.

68. Fattahi Z, Beheshtian M, Mohseni M, Poustchi H, Sellars E, Nezhadi SH, Amini A, Arzhangi S, Jalalvand K, Jamali P, Mohammadi Z, Davarnia B, Nikuei P, Oladnabi M, Mohammadzadeh A, Zohrehvand E, Nejatizadeh A, Shekari M, Bagherzadeh M, Shamsi-Gooshki E, Börno S, Timmermann B, Haghdoost A, Najafipour R, Khorram Khorshid HR, Kahrizi K, Malekzadeh R, Akbari MR, **Najmabadi H**. Iranome: A catalog of genomic variations in the Iranian population. Hum Mutat. 2019; 40(11):1968-84.

69. InanlooRahatloo K, Peymani F, Kahrizi K, **Najmabadi H**. Whole-transcriptome analysis reveals dysregulation of

actin-cytoskeleton pathway in intellectual disability patients. Neuroscience. 2019; 404:423-44.

70. Kariminejad A, Yazdan H, Rahimian E, Kalhor Z, Fattahi Z, Zonooz MF, **Najmabadi H**, Ashrafi M.SZT2 mutation in a boy with intellectual disability, seizures and autistic features. Eur J Med Genet. 2019; 62(9):103556.

71. Hosseini M, Fattahi Z, Abedini SS, Hu H, Ropers HH, Kalscheuer VM, **Najmabadi H**, Kahrizi K. GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. Am J Med Genet A. 2019; 179(1):13-9.

72. Mehregan H, Mohseni M, Jalalvand K, Arzhangi S, Nikzat N, Banihashemi S, Kahrizi K, **Najmabadi H**. Novel mutations in MYTH4-FERM domains of myosin 15 are associated with autosomal recessive nonsyndromic hearing loss. Int J Pediatr Otorhinolaryngol. 2019; 117:115-26.

73. Beheshtian M, Fattahi Z, Fadaee M, Vazehan R, Jamali P, Parsimehr E, Kamgar M, Zonooz MF, Mahdavi SS, Kalhor Z, Arzhangi S, Abedini SS, Kermani FS, Mojahedi F, Kalscheuer VM, Ropers HH, Kariminejad A, **Najmabadi H**, Kahrizi K. Identification of disease-causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clin Genet. 2019; 95(6):718-25.

74. Kahrizi K, Huber M, Galetzka D, Dewi S, Schröder J, Weis E, Kariminejad A, Fattahi Z, Ropers HH, Schweiger S, **Najmabadi H**, Winter J. Homozygous variants in the gene SCAPER cause syndromic intellectual disability. Am J Med Genet A. 2019; 179(7):1214-25.

75. Mehregan H, Mohseni M, Akbari M, Jalalvand K, Arzhangi S, Nikzat N, Kahrizi K, **Najmabadi H**. Novel mutations in KCNQ4, LHFPL5 and COCH genes in Iranian families with hearing impairment. Arch Iran Med. 2019; 22(4):189-197.

76. Najafi K, Gholami S, Moshtagh A, Bazrgar M, Sadatian N, Abbasi G, Rostami P, Khalili S, Babanejad M, Nourmohammadi B, Faramarzi Garous N, **Najmabadi H**, Kariminejad R. Chromosomal aberrations in pregnancy and fetal loss: Insight on the effect of consanguinity, review of 1625 cases. Mol Genet Genomic Med. 2019; 7(8):e820.

77. Babanejad M, Zarandy MM, Nikzat N, Bazazzadegan N, Arzhangi S, Mohseni M, Kahrizi K, **Najmabadi H**. G130V de novo mutation in an Iranian pedigree with nonsyndromic hearing loss without palmoplantar keratoderma. Int J Pediatr Otorhinolaryngol. 2019; 126:109607.

78. Ataei R, Khoshbakht S, Beheshtian M, Abedini SS, Behravan H, Esmaeili Dizghandi S, Godratpour F, Mirzaei S, Bahrami F, Akbari M, Keshavarzi F, Kahrizi K, **Najmabadi H**. Contribution of Iran in elucidating the genetic causes of autosomal recessive intellectual disability. Arch Iran Med. 2019; 22(8):461-71.

79. Bazazzadegan N, Vazehan R, Fadaee M, Fattahi Z, Abolhassani A, Parsimehr E, Kalhor Z, Faraji Zonooz M, Ahangari F, Dehdahsi S, Samiee F, Jamali P, Habibi H, Nourizadeh Y, Mahdavi S, Beheshtian M, Kariminejad A, Smith RJ, **Najmabadi H**. Brief report of variants detected in hereditary hearing loss cases in Iran over a 3-year period. Iran J Public Health. 2019; 48(10):1910-5.

80. Kariminejad A, Vahidnezhad H, Ghaderi-Sohi S, Ghannadan AR, Youssefian L, Parsimehr E, Faraji Zonooz M, Kariminejad MH, Uitto J, **Najmabadi H**, Hennekam RC. Widespread aplasia cutis congenita in sibs with PLEC1 and ITGB4 variants. Am J Med Genet A. 2019; 179(8):1547-55.

81. Mohammadzadeh A, Akbaroghli S, Aghaei-Moghadam E, Mahdieh N, Badv RS, Jamali P, Kariminejad R, Chavoshzadeh Z, Ghasemi Firouzabadi S, Mansour Ghanaie R, Nozari A, Banihashemi S, Hadipour F, Hadipour Z, Kariminejad A, **Najmabadi H**, Shafeghati Y, Behjati F. Investigation of chromosomal abnormalities and microdeletion/ microduplication(s) in fifty Iranian patients with multiple congenital anomalies. Cell J. 2019; 21(3):337-49.

82. Kariminejad A, Szenker-Ravi E, Lekszas C, Tajsharghi H, Moslemi AR, Naert T, Tran HT, Ahangari F, Rajaei M, Nasseri M, Haaf T, Azad A, Superti-Furga A, Maroofian R, Ghaderi-Sohi S, **Najmabadi H**, Abbaszadegan MR, Vleminckx K, Nikuei P, Reversade B. Homozygous null TBX4 mutations lead to posterior amelia with pelvic and

pulmonary hypoplasia. Am J Hum Genet. 2019; 105(6):1294-301.

83. Bekiesinska-Figatowska M, Hosseini M, Arzhangi S, **Najmabadi H**, Rosenfeld JA, Du H, Marafi D, Blaser S, Teitelbaum R, Silver R; Baylor-Hopkins Center for Mendelian Genomics, Posey JE, Ropers HH, Gibbs RA, Wiszniewski W, Lupski JR, Chitayat D, Kahrizi K, Gawlinski P. Bi-allelic pathogenic variants in TUBGCP2 cause microcephaly and lissencephaly spectrum disorders. Am J Hum Genet. 2019; 105(5):1005-15.

84. Palmer EE, Stuhlmann T, Weinert S, Haan E, Van Esch H, Holvoet M, Boyle J, Leffler M, Raynaud M, Moraine C, van Bokhoven H, Kleefstra T, Kahrizi K, **Najmabadi H**, Ropers HH, Delgado MR, Sirsi D, Golla S, Sommer A, Pietryga MP, Chung WK, Wynn J, Rohena L, Bernardo E, Hamlin D, Faux BM, Grange DK, Manwaring L, Tolmie J, Joss S; DDD Study, Cobben JM, Duijkers FAM, Goehringer JM, Challman TD, Hennig F, Fischer U, Grimme A, Suckow V, Musante L, Nicholl J, Shaw M, Lodh SP, Niu Z, Rosenfeld JA, Stankiewicz P, Jentsch TJ, Gecz J, Field M, Kalscheuer VM. De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Mol Psychiatry. 2018; 23(2):222-30.

85. Fattahi Z, Sheikh TI, Musante L, Rasheed M, Taskiran II, Harripaul R, Hu H, Kazeminasab S, Alam MR, Hosseini M, Larti F, Ghaderi Z, Celik A, Ayub M, Ansar M, Haddadi M, Wienker TF, Ropers HH, Kahrizi K, Vincent JB, **Najmabadi H**. Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Hum Mol Genet. 2018; 27(18):3177-88.

86. Abedini SS, Kahrizi K, de Pouplana LR, **Najmabadi H**. tRNA methyltransferase defects and intellectual disability. Arch Iran Med. 2018; 21(10):478-85.

87. Kazeminasab S, Taskiran II, Fattahi Z, Bazazzadegan N, Hosseini M, Rahimi M, Oladnabi M, Haddadi M, Celik A, Ropers HH, **Najmabadi H**, Kahrizi K. CNKSR1 gene defect can cause syndromic autosomal recessive intellectual disability. Am J Med Genet B Neuropsychiatr Genet. 2018; 177(8):691-9.

88. Abedini SS, Forouzesh Pour F, Karimi K, Ghaderi Z, Farashi S, Tavakoli Koudehi A, Javadi Pirouz H, Mobini Nejad SB, Azarkeivan A, **Najmabadi H**. Frequency of α-globin gene triplications and coinheritance with β-globin gene mutations in the Iranian population. Hemoglobin. 2018; 42(4):252-6.

89. Alavi A, Esmaeili S, Nafissi S, Kahrizi K, **Najmabadi H**. Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. Neuromuscul Disord. 2018; 28(4):303-14.

90. Babanejad M, Adeli OA, Nikzat N, Beheshtian M, Azarafra H, Sadeghnia F, Mohseni M, **Najmabadi H**, Kahrizi K. SLC52A2 mutations cause SCABD2 phenotype: A second report. Int J Pediatr Otorhinolaryngol. 2018; 104:195-9.

91. Booth KT, Azaiez H, Kahrizi K, Wang D, Zhang Y, Frees K, Nishimura C, **Najmabadi H**, Smith RJ. Exonic mutations and exon skipping: Lessons learned from DFNA5. Hum Mutat. 2018; 39(3):433-40.

92. Booth KT, Kahrizi K, Babanejad M, Daghagh H, Bademci G, Arzhangi S, Zareabdollahi D, Duman D, El-Amraoui A, Tekin M, **Najmabadi H**, Azaiez H, Smith RJ. Variants in CIB2 cause DFNB48 and not USH1J. Clin Genet. 2018; 93(4):812-21.

93. Booth KT, Kahrizi K, **Najmabadi H**, Azaiez H, Smith RJ. Old gene, new phenotype: splice-altering variants in CEACAM16 cause recessive non-syndromic hearing impairment. J Med Genet. 2018; 55(8):555-60.

94. Hosseini M, **Najmabadi H**, Kahrizi K. Calpains: Diverse functions but enigmatic. Arch Iran Med. 2018; 21(4):170-9.

95. Imtiaz A, Belyantseva IA, Beirl AJ, Fenollar-Ferrer C, Bashir R, Bukhari I, Bouzid A, Shaukat U, Azaiez H, Booth KT, Kahrizi K, **Najmabadi H**, Maqsood A, Wilson EA, Fitzgerald TS, Tlili A, Olszewski R, Lund M, Chaudhry T, Rehman AU, Starost MF, Waryah AM, Hoa M, Dong L, Morell RJ, Smith RJH, Riazuddin S, Masmoudi S, Kindt KS, Naz S, Friedman TB. CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Hum Mol

Genet. 2018; 27(5):780-98.

96. Kazeminasab S, **Najmabadi H**, Kahrizi K. Intellectual disability and ataxia: Genetic collisions. Arch Iran Med. 2018; 21(1):29-40.

97. Mehri M, Zarin M, Ardalani F, **Najmabadi H**, Azarkeivan A, Neishabury M. Novel mutations in mitochondrial carrier family gene SLC25A38, causing congenital sideroblastic anemia in Iranian families, identified by whole exome sequencing. Blood Cells Mol Dis. 2018; 71:39-44.

98. Nozari A, Aghaei-Moghadam E, Zeinaloo A, Mollazadeh R, Majnoon MT, Alavi A, Ghasemi Firouzabadi S, Mohammadzadeh A, Banihashemi S, Nikzaban M, **Najmabadi H**, Behjati F. A novel splicing variant in FLNC gene responsible for a highly penetrant familial dilated cardiomyopathy in an extended Iranian family. Gene. 2018; 659:160-7.

99. Charzewska A, Maiwald R, Kahrizi K, Oehl-Jaschkowitz B, Dufke A, Lemke JR, Enders H, **Najmabadi H**, Tzschach A, Hachmann W, Jensen C, Bienek M, Poznański J, Nawara M, Chilarska T, Obersztyn E, Hoffman-Zacharska D, Gos M, Bal J, Kalscheuer VM. The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12-related disorders. Clin Genet. 2018; 94(5):450-6.

100. Avenarius MR, Jung JY, Askew C, Jones SM, Hunker KL, Azaiez H, Rehman AU, Schraders M, **Najmabadi H**, Kremer H, Smith RJH, Géléoc GSG, Dolan DF, Raphael Y, Kohrman DC. Grxcr2 is required for stereocilia morphogenesis in the cochlea. PLoS One. 2018; 13(8):e0201713.

101. Fazeli Z, Ghaderian SMH, **Najmabadi H**, Omrani MD. High expression of miR-510 was associated with CGG expansion located at upstream of FMR1 into full mutation. J Cell Biochem. 2018. doi: 10.1002/jcb.27505. Online ahead of print.PMID: 30160796

102. Michel V, Booth KT, Patni P, Cortese M, Azaiez H, Bahloul A, Kahrizi K, Labbé M, Emptoz A, Lelli A, Dégardin J, Dupont T, Aghaie A, Oficjalska-Pham D, Picaud S, **Najmabadi H**, Smith RJ, Bowl MR, Brown SD, Avan P, Petit C, El-Amraoui A. CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Mol Med. 2017; 9(12):1711-31.

103. Kariminejad A, Dahl-Halvarsson M, Ravenscroft G, Afroozan F, Keshavarz E, Goullée H, Davis MR, Faraji Zonooz M, **Najmabadi H**, Laing NG, Tajsharghi H. TOR1A variants cause a severe arthrogryposis with developmental delay, strabismus and tremor. Brain. 2017; 140(11):2851-9.

104. Edizadeh M, Vazehan R, Javadi F, Dehdahsi S, Fadaee M, Faraji Zonooz M, Parsimehr E, Ahangari F, Abolhassani A, Kalhor Z, Fattahi Z, Beheshtian M, Kariminejad A, Akbari MR, **Najmabadi H**, Nafissi S. De novo Mutation in CACNA1S Gene in a 20-Year-Old Man Diagnosed with Metabolic Myopathy. Arch Iran Med. 2017; 20(9):617-20.

105. Vermeulen C, Geeven G, de Wit E, Verstegen MJAM, Jansen RPM, van Kranenburg M, de Bruijn E, Pulit SL, Kruisselbrink E, Shahsavari Z, Omrani D, Zeinali F, **Najmabadi H**, Katsila T, Vrettou C, Patrinos GP, Traeger-Synodinos J, Splinter E, Beekman JM, Kheradmand Kia S, Te Meerman GJ, Ploos van Amstel HK, de Laat W. Sensitive Monogenic Noninvasive Prenatal Diagnosis by Targeted Haplotyping. Am J Hum Genet. 2017; 101(3):326-39.

106. Ivanova EL, Mau-Them FT, Riazuddin S, Kahrizi K, Laugel V, Schaefer E, de Saint Martin A, Runge K, Iqbal Z, Spitz MA, Laura M, Drouot N, Gérard B, Deleuze JF, de Brouwer APM, Razzaq A, Dollfus H, Assir MZ, Nitchké P, Hinckelmann MV, Ropers H, Riazuddin S, **Najmabadi H**, van Bokhoven H, Chelly J. Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. Am J Hum Genet. 2017; 101(3):428-40.

107. Alavi A, Esmaeili S, Nilipour Y, Nafissi S, Tonekaboni SH, Zamani G, Ashrafi MR, Kahrizi K, **Najmabadi H**, Jazayeri F. LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. J Neurogenet. 2017;

31(3):161-9.

108. Alber M, Kalscheuer VM, Marco E, Sherr E, Lesca G, Till M, Gradek G, Wiesener A, Korenke C, Mercier S, Becker F, Yamamoto T, Scherer SW, Marshall CR, Walker S, Dutta UR, Dalal AB, Suckow V, Jamali P, Kahrizi K, **Najmabadi H**, Minassian BA. ARHGEF9 disease: Phenotype clarification and genotype-phenotype correlation. Neurol Genet. 2017; 3(3):e148.

109. 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; 38(6):621-36.

110. Amos JS, Huang L, Thevenon J, Kariminedjad A, Beaulieu CL, Masurel-Paulet A, **Najmabadi H**, Fattahi Z, Beheshtian M, Tonekaboni SH, Tang S, Helbig KL, Alcaraz W, Rivière JB, Faivre L, Innes AM, Lebel RR, Boycott KM. Autosomal recessive mutations in THOC6 cause intellectual disability: Syndrome delineation requiring forward and reverse phenotyping. Clin Genet. 2017; 91(1):92-9.

111. Fattahi Z, Kalhor Z, Fadaee M, Vazehan R, Parsimehr E, Abolhassani A, Beheshtian M, Zamani G, Nafissi S, Nilipour Y, Akbari MR, Kahrizi K, Kariminejad A, **Najmabadi H**. Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. Clin Genet. 2017; 91(3):386-402.

112. Firouzabadi SG, Kariminejad R, Vameghi R, Darvish H, Ghaedi H, Banihashemi S, Firouzkouhi Moghaddam M, Jamali P, Mofidi Tehrani HF, Dehghani H, Narooie-Nejad M, Jamshidi J, Tafakhori A, Sadabadi S, **Najmabadi H**, Behjati F. Copy Number Variants in Patients with Autism and Additional Clinical Features: Report of VIPR2 Duplication and a Novel Microduplication Syndrome. Mol Neurobiol. 2017; 54(9):7019-27.

113. Farashi S, Rad F, Shahmohammadi B, Imanian H, Azarkeivan A, **Najmabadi H**. First report of a dominantly inherited β-thalassemia caused by a novel elongated β-globin chain. Hemoglobin. 2016; 40(2):102-7.

114. Beheshtian M, Izadi N, Kriegshauser G, Kahrizi K, Mehr EP, Rostami M, Hosseini M, Azad M, Montajabiniat M, Kariminejad A, Nemeth S, Oberkanins C, **Najmabadi H**. Prevalence of common MEFV mutations and carrier frequencies in a large cohort of Iranian populations. J Genet. 2016; 95(3):667-74.

115. Beheshtian M, Babanejad M, Azaiez H, Bazazzadegan N, Kolbe D, Sloan-Heggen C, Arzhangi S, Booth K, Mohseni M, Frees K, Azizi MH, Daneshi A, Farhadi M, Kahrizi K, Smith RJ, **Najmabadi H**. Heterogeneity of hereditary hearing loss in Iran: A comprehensive review. Arch Iran Med. 2016; 19(10):720-8.

116. Mohseni M, Razzaghmanesh M, Parsi Mehr E, Zare H, Beheshtian M, **Najmabadi H**. Novel CFTR mutations in two Iranian families with severe cystic fibrosis. Iran Biomed J. 2016; 20(4):201-6.

117. Babanejad M, Moein H, Akbari MR, Badiei A, Yaseri M, Soheilian M, **Najmabadi H**. Investigating the CFH gene polymorphisms as a risk factor for age-related macular degeneration in an Iranian population. Ophthalmic Genet. 2016; 37(2):144-9.

118. Mehregan H, **Najmabadi H**, Kahrizi K. Genetic studies in intellectual disability and behavioral impairment. Arch Iran Med. 2016; 19(5):363-75.

119. Esmaeeli-Nieh S, Fenckova M, Porter IM, Motazacker MM, Nijhof B, Castells-Nobau A, Asztalos Z, Weißmann R, Behjati F, Tzschach A, Felbor U, Scherthan H, Sayfati SM, Ropers HH, Kahrizi K, **Najmabadi H**, Swedlow JR, Schenck A, Kuss AW. BOD1 is required for cognitive function in humans and drosophila. PLoS Genet. 2016; 12(5):e1006022.

120. Esmail Nia G, Fadaee M, Royer R, **Najmabadi H**, Akbari MR. Profiling Fanconi Anemia Gene Mutations among Iranian Patients. Arch Iran Med. 2016; 19(4):236-40.

121. Fadaee M, Kariminejad A, Fattahi Z, Nafissi S, Godarzi HR, Beheshtian M, Vazehan R, Akbari MR, Kahrizi K, **Najmabadi H**. Report of limb girdle muscular dystrophy type 2a in 6 Iranian patients, one with a novel deletion in CAPN3 gene. Neuromuscul Disord. 2016; 26 (4-5):277-82.

122. Faraji Zonooz M, Sabbagh-Kermani F, Fattahi Z, Fadaee M, Akbari MR, Amiri R, Vahidnezhad H, Uitto J, **Najmabadi H**, Kariminejad A. Whole genome linkage analysis followed by whole exome sequencing identifies Nicastrin (NCSTN) as a causative gene in a multiplex family with γ-secretase spectrum of autoinflammatory skin phenotypes. J Invest Dermatol. 2016; 136(6):1283-6.

123. Farashi S, Rad F, Shahmohammadi B, Imanian H, Azarkeivan A, **Najmabadi H**. First report of a dominantly inherited β-thalassemia caused by a novel elongated β-globin Chain. Hemoglobin. 2016; 40(2):102-7.

124. Farashi S, Garous NF, Vakili S, Ashki M, Imanian H, Azarkeivan A, **Najmabadi H**. Characterization of Homozygous Hb Setif (HBA2: c.283G>T) in the Iranian Population. Hemoglobin. 2016; 40(1):53-5.

125. Farashi S, Vakili S, Garous NF, Ashki M, Forouzesh Pour F, Zeinali F, Rad F, Imanian H, Azarkeivan A, **Najmabadi H**. Identification of Mutations Causing Aberrant Termination and Deficient Splice Donor Site on the HBA1 Gene. Hemoglobin. 2016; 40(1):38-43

126. Farashi S, Bayat N, Vakili S, Faramarzi Garous N, Ashki M, Imanian H, **Najmabadi H**, Azarkeivan A. Point mutations which should not be overlooked in Hb H disease. Expert Rev Hematol. 2016; 9(1):107-13.

127. 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; 24(3):392-9.

128. Adegbola A, Musante L, Callewaert B, Maciel P, Hu H, Isidor B, Picker-Minh S, Le Caignec C, Delle Chiaie B, Vanakker O, Menten B, Dheedene A, Bockaert N, Roelens F, Decaestecker K, Silva J, Soares G, Lopes F, **Najmabadi H**, Kahrizi K, Cox GF, Angus SP, Staropoli JF, Fischer U, Suckow V, Bartsch O, Chess A, Ropers HH, Wienker TF, Hübner C, Kaindl AM, Kalscheuer VM. Redefining the MED13L syndrome. Eur J Hum Genet. 2015; 23(10):1308-17.

129. Beheshtian M, Saee Rad S, Babanejad M, Mohseni M, Hashemi H, Eshghabadi A, Hajizadeh F, Akbari MR, Kahrizi K, Riazi Esfahani M, **Najmabadi H**. Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. Arch Iran Med. 2015; 18(11):776-85.

130. 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; 23(3):331-6.

131. Kashef A, Nikzat N, Bazzazadegan N, Fattahi Z, Sabbagh-Kermani F, Taghdiri M, Azadeh B, Mojahedi F, Khoshaeen A, Habibi H, **Najmabadi H**, Kahrizi K. Finding mutation within non-coding region of GJB2 reveals its importance in genetic testing of hearing loss in Iranian population. Int J Pediatr Otorhinolaryngol. 2015; 79(2):136-8.

132. Farashi S, Garous NF, Ashki M, Vakili S, Zeinali F, Imanian H, Azarkeivan A, Giordano PC, **Najmabadi H**. Homozygosity for the AATAAA > AATA- - Polyadenylation Site Mutation on the α2-Globin Gene Causing Transfusion-Dependent Hb H Disease in an Iranian Patient: A Case Report. Hemoglobin. 2015; 39(5):355-8.

133. Farashi S, Vakili S, Garous NF, Ashki M, Imanian H, Azarkeivan A, **Najmabadi H**. Mutations on the α2-Globin Gene That May Trigger α(+)-Thalassemia. Hemoglobin. 2015; 39(6):398-402.

134. Farashi S, Vakili S, Faramarzi Garous N, Ashki M, Imanian H, Azarkeivan A, **Najmabadi H**. Copy number variations of six and seven α-globin genes in a family with intermedia and major thalassemia phenotypes. Expert Rev Hematol. 2015; 8(5):693-8.

135. Farashi S, **Najmabadi H**. Diagnostic pitfalls of less well recognized HbH disease. Blood Cells Mol Dis. 2015; 55(4):387-95.

136. Farashi S, Faramarzi Garous N, Ashki M, Vakili S, Zeinali F, Imanian H, Azarkeivan A, **Najmabadi H**. Hb Dartmouth (HBA2: c.200T>C): An α2-globin gene associated with Hb H disease in one homozygous patient. Hemoglobin. 2015; 39(3):152-5.

137. **Farashi S**, **Faramarzi Garous N**, **Zeinali F**, **Vakili S**, **Ashki M**, **Imanian H**, **Najmabadi H**, **Azarkeivan A**, **Tamaddoni A**. A 21 nucleotide duplication on the α1- and α2-globin genes involves a variety of hypochromic microcytic anemias, from mild to Hb H disease. **Hemoglobin. 2015;** 39(3):196-200.

138. Farashi S, Bayat N, Faramarzi Garous N, Ashki M, Montajabi Niat M, Vakili S, Imanian H, Zeinali S, **Najmabadi H**, Azarkeivan A. Interaction of an α-globin gene triplication with β-globin gene mutations in Iranian patients with β-thalassemia intermedia. Hemoglobin. 2015; 39(3):201-6.

139. 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, Mozhdehipanah H, Rashidinejad A, Samiei S, Ghadami M, Windpassinger C, Gillessen-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; 24(20):5697-710.

140. Fattahi Z, Kahrizi K, Nafissi S, Fadaee M, Abedini SS, Kariminejad A, Akbari MR, **Najmabadi H**. Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. Arch Iran Med. 2015; 18(1):60-4.

141. Booth KT, Azaiez H, Kahrizi K, Simpson AC, Tollefson WT, Sloan CM, Meyer NC, Babanejad M, Ardalani F, Arzhangi S, Schnieders MJ, **Najmabadi H**, Smith RJ. PDZD7 and hearing loss: More than just a modifier.Am J Med Genet A. 2015; 167A(12):2957-65.

142. Mehrjoo Z, Babanejad M, Kahrizi K, **Najmabadi H**. Two novel mutations in ILDR1 gene cause autosomal recessive nonsyndromic hearing loss in consanguineous Iranian families. J Genet. 2015; 94(3):483-7.

143. Mehrjoo Z, Akbari MR, Abedini SS, Vaziri S, Kahrizi K, **Najmabadi H**. Carrier testing in known autosomal recessive intellectual disability genes in an Iranian healthy individual using exome sequencing. Arch Iran Med. 2015; 18(10):643-69.

144. Jazayeri R, Hu H, Fattahi Z, Musante L, Abedini SS, Hosseini M, Wienker TF, Ropers HH, **Najmabadi H**, Kahrizi K. Exome sequencing and linkage analysis identified novel candidate genes in recessive intellectual disability associated with ataxia. Arch Iran Med. 2015; 18(10):670-82.

145. Akhtarkhavari T, Joghataei MT, Fattahi Z, Akbari MR, Larti F, **Najmabadi H**, Kahrizi K. Genetic investigation of an Iranian supercentenarian by whole exome sequencing. Arch Iran Med. 2015; 18(10):688-97.

146. Sloan-Heggen CM, Babanejad M, Beheshtian M, Simpson AC, Booth KT, Ardalani F, Frees KL, Mohseni M, Mozafari R, Mehrjoo Z, Jamali L, Vaziri S, Akhtarkhavari T, Bazazzadegan N, Nikzat N, Arzhangi S, Sabbagh F, Otukesh H, Seifati SM, Khodaei H, Taghdiri M, Meyer NC, Daneshi A, Farhadi M, Kahrizi K, Smith RJ, Azaiez H, **Najmabadi H**. Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. J Med Genet. 2015; 52(12):823-9.

147. Beheshtian M, Saee Rad S, Babanejad M, Mohseni M, Hashemi H, Eshghabadi A, Hajizadeh F, Akbari MR, Kahrizi K, Riazi Esfahani M, **Najmabadi H**. Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. Arch Iran Med. 2015; 18(11):776-85.

148. Davarniya B, Hu H, Kahrizi K, Musante L, Fattahi Z, Hosseini M, Maqsoud F, Farajollahi R, Wienker TF, Ropers

HH, **Najmabadi H**.The role of a novel TRMT1 gene mutation and rare GRM1 gene defect in intellectual disability in two Azeri families. PLoS One. 2015; 10(8):e0129631.

149. Mehrjoo Z, Najmabadi A, Abedini SS, Mohseni M, Kamali K, **Najmabadi H**, Khorram Khorshid HR. Association study of the TREM2 gene and identification of a novel variant in exon 2 in Iranian patients with late-onset Alzheimer's disease. Med Princ Pract. 2015; 24(4):351-4.

150. Rostami P, Valizadegan S, Ghalandary M, Mehrjouy MM, Esmail-Nia G, Khalili S, Shahmoradi SS, Imanian H, Hadavi V, Ghaderi-Sohi S, Almadani N, Afroozan F, Kariminejad A, Kariminejad R, **Najmabadi H**. Prenatal Screening for Aneuploidies Using QF-PCR and Karyotyping: A Comprehensive Study in Iranian Population. Arch Iran Med. 2015; 18(5):296-303.

151. Oladnabi M, Musante L, Larti F, Hu H, Abedini SS, Wienker T, Ropers HH, Kahrizi K, **Najmabadi H**. New evidence for the role of calpain 10 in autosomal recessive intellectual disability: identification of two novel nonsense variants by exome sequencing in Iranian families. Arch Iran Med. 2015; 18(3):179-84.

152. Iqbal Z, Willemsen MH, Papon MA, Musante L, Benevento M, Hu H, Venselaar H, Wissink-Lindhout WM, Vulto-van Silfhout AT, Vissers LE, de Brouwer AP, Marouillat S, Wienker TF, Ropers HH, Kahrizi K, Nadif Kasri N, **Najmabadi H**, Laumonnier F, Kleefstra T, van Bokhoven H. Homozygous SLC6A17 mutations cause autosomal-recessive intellectual disability with progressive tremor, speech impairment, and behavioral problems. Am J Hum Genet. 2015; 96(3):386-96.

153. Zaker-Kandjani B, Namdar-Aligoodarzi P, Azarkeivan A, **Najmabadi H**, Banan M. Mutation screening of the Krüppel-like factor 1 gene using single-strand conformational polymorphism in a cohort of Iranian β-thalassemia patients. Hemoglobin. 2015; 39(1):24-9.

154. Taghizadeh S, **Najmabadi H**, Kamali K, Behjati F. Evaluation of chromosomal aberrations caused by air pollutants in some taxi drivers from two polluted districts of urban Tehran and its comparison with drivers from rural areas of Lahijan: a pilot study. J Environ Health Sci Eng. 2014; 12(1):144.

155. Shearer AE, Eppsteiner RW, Booth KT, Ephraim SS, Gurrola J, Simpson A, Black-Ziegelbein EA, Joshi S, Ravi H, Giuffre AC, Happe S, Hildebrand MS, Azaiez H, Bayazit YA, Erdal ME, Lopez-Escamez JA, Gazquez I, Tamayo ML, Gelvez NY, Leal GL, Jalas C, Ekstein J, Yang T, Usami S, Kahrizi K, Bazazzadegan N, **Najmabadi H**, Scheetz TE, Braun TA, Casavant TL, LeProust EM, Smith RJ. Utilizing ethnic-specific differences in minor allele frequency to recategorize reported pathogenic deafness variants. Am J Hum Genet. 2014;95(4):445-53.

156. **Najmabadi H**, Kahrizi K. Genetics of non-syndromic hearing loss in the Middle East. Int J Pediatr Otorhinolaryngol. 2014;78(12):2026-36. Poopak B, Saki N, Purfatholah AA, **Najmabadi H**, Mortazavi Y, Arzanian MT, Khosravipour G, Haghnejad F, Salari F, Shahjahani M. Pattern of immunoglobulin and T-cell receptor-δ/γ gene rearrangements in Iranian children with B-precursor acute lymphoblastic leukemia. Hematology. 2014;19(5):259-66.

157. Mohseni M, Honarpour A, Mozafari R, Davarnia B, **Najmabadi H**, Kahrizi K. Identification of a founder mutation for Pendred syndrome in families from northwest Iran. Int J Pediatr Otorhinolaryngol. 2014; 78(11):1828-32.

158. Hu H, Wienker TF, Musante L, Kalscheuer VM, Kahrizi K, **Najmabadi H**, Ropers HH. Integrated sequence analysis pipeline provides one-stop solution for identifying disease-causing mutations. Hum Mutat. 2014; 35(12):1427-35.

159. Reuter MS, Musante L, Hu H, Diederich S, Sticht H, Ekici AB, Uebe S, Wienker TF, Bartsch O, Zechner U, Oppitz C, Keleman K, Jamra RA, **Najmabadi H**, Schweiger S, Reis A, Kahrizi K. NDST1 missense mutations in autosomal recessive intellectual disability. Am J Med Genet A. 2014; 164A(11):2753-63.

160. Kariminejad A, Bozorgmehr B, Najafi A, Khoshaeen A, Ghalandari M, **Najmabadi H**, Kariminejad MH, Vanakker OM, Hosen MJ, Malfait F, Quaglino D, Florijn RJ, Bergen AA, Hennekam RC. Retinitis pigmentosa, cutis laxa, and pseudoxanthoma elasticum-like skin manifestations associated with GGCX mutations. J Invest Dermatol. 2014;

134(9):2331-8.

161. Loghmani Khouzani H, Kariminejad A, Zamani G, Ghalandary M, Bozorgmehr B, Amirsalari S, Mojahedi F, Tonekaboni SH, Kariminejad R, **Najmabadi H**. Investigation of microdeletions in syndromic intellectual disability by MLPA in Iranian population. Arch Iran Med. 2014; 17(7):471-4.

162. Jamaldini SH, Babanejad M, Mozaffari R, Nikzat N, Jalalvand K, Badiei A, Sanati H, Shakerian F, Afshari M, Kahrizi K, **Najmabadi H**. Association of polymorphisms at LDLR locus with coronary artery disease independently from lipid profile. Acta Med Iran. 2014; 52(5):352-9.

163. Pazhoomand R, Keyhani E, Banan M, **Najmabadi H**, Khodadadi F, Iraniparast A, Feiz F, Majidzadeh K, Bahman I, Moghadam FA, Sobhani AM, Muhammadnejad A, Abedini SS, Behjati F. Detection of HER2 status in breast cancer: comparison of current methods with MLPA and real-time RT-PCR. Asian Pac J Cancer Prev. 2013;14(12):7621-8.

164. Behjati F, Firouzabadi SG, Kariminejad R, Vameghi R, Sajedi F, Shafaghati Y, Ghasemlou BE, Shojaei A, Jamali P, Bahman I, **Najmabadi H**. Genomic characterization of some Iranian children with idiopathic mental retardation using array comparative genomic hybridization. Indian J Hum Genet. 2013;19(4):443-8.

165. Behjati F, Ghasemi Firouzabadi S, Sajedi F, Kahrizi K, Najafi M, Ebrahimizade Ghasemlou B, Shafeghati Y, Behnia F, Mohammadi Arya AR, Karimi H, Hadipour F, Hadipour Z, Jamali P, Kariminejad R, Darvish H, Bahman I, Bagherizadeh E, **Najmabadi H**, Vameghi R. Identification of Chromosome Abnormalities in Subtelomeric Regions Using Multiplex Ligation Dependent Probe Amplification (MLPA) Technique in 100 Iranian Patients With Idiopathic Mental Retardation. Iran Red Crescent Med J. 2013; 15(10):e8221.

166. Akbariazar E, Ebrahimpour M, Akbari S, Arzhanghi S, Abedini SS, **Najmabadi H**, Kahrizi K. A novel deletion mutation in ASPM gene in an Iranian family with autosomal recessive primary microcephaly. Iran J Child Neurol. 2013; 7(2):23-30.

167. Bayat N, Farashi S, Hafezi-Nejad N, Faramarzi N, Ashki M, Vakili S, Imanian H, Khosravi M, Azar-Keivan A, **Najmabadi H**. Novel mutations responsible for α-thalassemia in Iranian families. Hemoglobin. 2013; 37(2):148-59.

168. Hafezi-Nejad N, Khosravi M, Bayat N, Kariminejad A, Hadavi V, Oberkanins C, Azarkeivan A, **Najmabadi H**. Characterizing a cohort of α-thalassemia couples collected during screening for hemoglobinopathies: 14 Years of an Iranian Experience. Hemoglobin. 2014; 38(3):153-7.

169. Sahraian S, Babashams M, Reza-Soltani P, **Najmabadi H**, Kahrizi K, Gorgani SH. Serotonin transporter polymorphism (5-HTTLPR) and citalopram effectiveness in Iranian patients with major depressive disorder. Iran J Psychiatry. 2013; 8(2):86-91.

170. Ben Ammar A, Soltanzadeh P, Bauché S, Richard P, Goillot E, Herbst R, Gaudon K, Huzé C, Schaeffer L, Yamanashi Y, Higuchi O, Taly A, Koenig J, Leroy JP, Hentati F, **Najmabadi H**, Kahrizi K, Ilkhani M, Fardeau M, Eymard B, Hantaï D. A mutation causes MuSK reduced sensitivity to agrin and congenital myasthenia. PLoS One. 2013; 8(1):e53826.

171. Banan M, Bayat H, Namdar-Aligoodarzi P, Azarkeivan A, Kamali K, Daneshmand P, Zaker-Kandjani B, **Najmabadi H**. Utility of the multivariate approach In predicting β-thalassemia intermedia or β-thalassemia major types In Iranian patients. Hemoglobin. 2013; 37(5):413-22.

172. Neishabury M, Zamani F, Keyhani E, Azarkeivan A, Abedini SS, Eslami MS, Kakroodi ST, Vesiehsari MJ, **Najmabadi H**. The influence of the BCL11A polymorphism on the phenotype of patients with beta thalassemia could be affected by the beta globin locus control region and/or the Xmn1-HBG2 genotypic background. Blood Cells Mol Dis. 2013; 51(2):80-4.

173. Bayat N, Farashi S, Hafezi-Nejad N, Faramarzi N, Ashki M, Vakili S, Imanian H, Khosravi M, Azar-Keivan A,

Najmabadi H. Novel mutations responsible for α-thalassemia in Iranian families. Hemoglobin. 2013; 37(2):148-59.

174. Pourfarzad F, von Lindern M, Azarkeivan A, Hou J, Kia SK, Esteghamat F, van Ijcken W, Philipsen S, **Najmabadi H**, Grosveld F. Hydroxyurea responsiveness in β-thalassemic patients is determined by the stress response adaptation of erythroid progenitors and their differentiation propensity. Haematologica. 2013; 98(5):696-704.

175. 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; 161A(8):1915-22.

176. Al-Allawi NA, Jalal SD, Rasheed NS, Bayat N, Imanian H, **Najmabadi H**, Faraj A. The spectrum of α-thalassemia mutations in the Kurdish population of Northeastern Iraq. Hemoglobin. 2013; 37(1):56-64.

177. Zeinali F, Mohseni M, Fadaee M, Fattahi Z, **Najmabadi H**, Otukesh H, Kahrizi K. Investigation of ATP6V1B1 and ATP6V0A4 genes causing hereditary hearing loss associated with distal renal tubular acidosis in Iranian families. J Laryngol Otol. 2014; 128(12):1056-9.

178. Kahrizi K, Bazazzadegan N, Jamali L, Nikzat N, Kashef A, **Najmabadi H**. A novel mutation of the USH2C (GPR98) gene in an Iranian family with Usher syndrome type II. J Genet. 2014; 93(3):837-41.

179. Fattahi Z, Rostami P, Najmabadi A, Mohseni M, Kahrizi K, Akbari MR, Kariminejad A, **Najmabadi H**. Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of eight novel mutations in five BBS gene. J Hum Genet. 2014; 59(7):368-75.

180. Davarnia B, Babanejad M, Fattahi Z, Nikzat N, Bazazzadegan N, Pirzade A, Farajollahi R, Nishimura C, Jalalvand K, Arzhangi S, Kahrizi K, Smith RJ, **Najmabadi H**. Spectrum of GJB2 (Cx26) gene mutations in Iranian Azeri patients with nonsyndromic autosomal recessive hearing loss. Int J Pediatr Otorhinolaryngol. 2012; 76(2):268-71.

181. Optimizing A Lipocomplex-Based Gene Transfer Method into HeLa Cell Line. Asgharian A, Banan M, **Najmabadi H**. Optimizing a lipocomplex-based gene transfer method into HeLa cell line. Cell J. 2014; 15(4):372-7.

182. Khatami S, **Najmabadi H**, Rouhi S, Mirzazadeh R, Bayat P, Sadeghi S. Preliminary identification of hemoglobin q-iran in an Iranian family from central province of Iran by globin chain analysis on HPLC. Arch Iran Med. 2013; 16(12):739-40.

183. Banan M, Bayat H, Namdar-Aligoodarzi P, Azarkeivan A, Kamali K, Daneshmand P, Zaker-Kandjani B, **Najmabadi H**. Utility of the multivariate approach In predicting β-thalassemia intermedia or β-thalassemia major types In Iranian patients. Hemoglobin. 2013; 37(5):413-22.

184. Pourfarzad F, von Lindern M, Azarkeivan A, Hou J, Kia SK, Esteghamat F, van Ijcken W, Philipsen S, **Najmabadi H**, Grosveld F. Hydroxyurea responsiveness in β-thalassemic patients is determined by the stress response adaptation of erythroid progenitors and their differentiation propensity. Haematologica. 2013; 98(5):696-704.

185. Sahraian S, Babashams M, Reza-Soltani P, **Najmabadi H**, Kahrizi K, Gorgani SH. Serotonin Transporter Polymorphism (5-HTTLPR) and Citalopram Effectiveness in Iranian Patients with Major Depressive Disorder. Iran J Psychiatry. 2013; 8(2):86-91.

186. Jazayeri R, Qoreishi M, Hoseinzadeh HR, Babanejad M, Bakhshi E, **Najmabadi H**, Jazayeri SM. Investigation of the asporin gene polymorphism as a risk factor for knee osteoarthritis in Iran. Am J Orthop (Belle Mead NJ). 2013; 42(7):313-6.

187. Ben Ammar A, Soltanzadeh P, Bauché S, Richard P, Goillot E, Herbst R, Gaudon K, Huzé C, Schaeffer L, Yamanashi Y, Higuchi O, Taly A, Koenig J, Leroy JP, Hentati F, **Najmabadi H**, Kahrizi K, Ilkhani M, Fardeau M, Eymard B, Hantaï D. A mutation causes MuSK reduced sensitivity to agrin and congenital myasthenia. PLoS One. 2013;

8(1):e53826.

188. Bayat N, Farashi S, Hafezi-Nejad N, Faramarzi N, Ashki M, Vakili S, Imanian H, Khosravi M, Azar-Keivan A, **Najmabadi H**. Novel mutations responsible for α-thalassemia in Iranian families. Hemoglobin. 2013; 37(2):148-59.

189. Pourfarzad F, von Lindern M, Azarkeivan A, Hou J, Kia SK, Esteghamat F, van Ijcken W, Philipsen S, **Najmabadi H**, Grosveld F. Hydroxyurea responsiveness in β-thalassemic patients is determined by the stress response adaptation of erythroid progenitors and their differentiation propensity. Haematologica. 2013; 98(5):696-704.

190. Neishabury M, Zamani F, Keyhani E, Azarkeivan A, Abedini SS, Eslami MS, Kakroodi ST, Vesiehsari MJ, **Najmabadi H**. The influence of the BCL11A polymorphism on the phenotype of patients with beta thalassemia could be affected by the beta globin locus control region and/or the Xmn1-HBG2 genotypic background. Blood Cells Mol Dis. 2013; 51(2):80-4.

191. 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; 161(8):1915-22.

192. Al-Allawi NA, Jalal SD, Rasheed NS, Bayat N, Imanian H, **Najmabadi H**, Faraj A. The spectrum of α-thalassemia mutations in the Kurdish population of Northeastern Iraq. Hemoglobin. 2013; 37(1):56-64.

193. Fattahi Z, **Najmabadi H**. Prevalence of ACTN3 (the athlete gene) R577X polymorphism in Iranian population. Iran Red Crescent Med J. 2012; 14(10):617-22.

194. Banan M, Esmaeilzadeh-Gharehdaghi E, Nezami M, Deilami Z, Farashi S, Philipsen S, Esteghamat F, Pourfarzad F, Ali Imam AM, **Najmabadi H**. cAMP response element-binding protein 1 is required for hydroxyurea-mediated induction of γ-globin expression in K562 cells. Clin Exp Pharmacol Physiol. 2012; 39(6):510-7.

195. Soltani Banavandi MJ, Kahrizi K, Behjati F, Mohseni M, Darvish H, Bahman I, Abedinni SS, Ghasemi Firouzabadi S, Jafari E, Ghadami Sh, Sabbagh F, Kavoosi GR, **Najmabadi H**. Investigation of genetic causes of intellectual disability in Kerman province, South East of Iran. Iran Red Crescent Med J. 2012; 14(2):79-85.

196. 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; 83(5):488-90.

197. Hosseini MM, Tonekaboni SH, Papari E, Bahman I, Behjati F, Kahrizi K, **Najmabadi H**. A novel mutation in MCPH1 gene in an Iranian family with primary microcephaly. J Pak Med Assoc. 2012; 62(11):1244-7.

198. Babanejad M, Fattahi Z, Bazazzadegan N, Nishimura C, Meyer N, Nikzat N, Sohrabi E, Najmabadi A, Jamali P, Habibi F, Smith RJ, Kahrizi K, **Najmabadi H**. A comprehensive study to determine heterogeneity of autosomal recessive nonsyndromic hearing loss in Iran. Am J Med Genet A. 2012; 158A(10):2485-92.

199. Botla SK, Gholami AM, Malekpour M, Moskalev EA, Fallah M, Jandaghi P, Aghajani A, Bondar IS, Omranipour R, Malekpour F, Mohajeri A, Babadi AJ, Sahin Ö, Bubnov VV, **Najmabadi H**, Hoheisel JD, Riazalhosseini Y. Diagnostic values of GHSR DNA methylation pattern in breast cancer. Breast Cancer Res Treat. 2012; 135(3):705-13.

200. Zamani F, Bagheri Z, Bayat M, Fereshtehnejad SM, Basi A, **Najmabadi H**, Ajdarkosh H. Iranian hereditary hemochromatosis patients: Baseline characteristics, laboratory data and gene mutations. Med Sci Monit. 2012; 18(10):CR622-629.

201. Davarnia B, Babanejad M, Fattahi Z, Nikzat N, Bazazzadegan N, Pirzade A, Farajollahi R, Nishimura C, Jalalvand K, Arzhangi S, Kahrizi K, Smith RJ, **Najmabadi H**. Spectrum of GJB2 (Cx26) gene mutations in Iranian Azeri patients with nonsyndromic autosomal recessive hearing loss. Int J Pediatr Otorhinolaryngol. 2012; 76(2):268-71.

202. Banan M, Bayat H, Azarkeivan A, Mohammadparast S, Kamali K, Farashi S, Bayat N, Khani MH, Neishabury M, **Najmabadi H**. The XmnI and BCL11A single nucleotide polymorphisms may help predict hydroxyurea response in Iranian β-thalassemia patients. Hemoglobin. 2012; 36(4):371-80.

203. Fattahi Z, Shearer AE, Babanejad M, Bazazzadegan N, Almadani SN, Nikzat N, Jalalvand K, Arzhangi S, Esteghamat F, Abtahi R, Azadeh B, Smith RJ, Kahrizi K, **Najmabadi H**. Screening for MYO15A gene mutations in autosomal recessive nonsyndromic, GJB2 negative Iranian deaf population. Am J Med Genet A. 2012; 158A(8):1857-64.

204. Mahdavi MR, Bayat N, Hadavi V, Karami H, Roshan P, **Najmabadi H**, Rohanizadeh H. Report of haemoglobin J-Toronto and alpha thalassemia in a family from North of Iran. J Pak Med Assoc. 2012; 62(4):396-8.

205. Abedini SS, Kahrizi K, Behjati F, Banihashemi S, Ghasemi Firoozabadi S, **Najmabadi H**. Mutational Screening of ARX Gene in Iranian Families with X-linked Intellectual Disability. Arch Iran Med. 2012; 15(6):361-5.

206. Soltani Banavandi MJ, Kahrizi K, Behjati F, Mohseni M, Darvish H, Bahman I, Abedinni SS, Ghasemi Firouzabadi S, Jafari E, Ghadami Sh, Sabbagh F, Kavoosi GR, **Najmabadi H**. Investigation of genetic causes of intellectual disability in Kerman province, South East of Iran. Iran Red Crescent Med J. 2012; 14(2):79-85.

207. Heidari A, Nariman Saleh Fam Z, Esmaeilzadeh-Gharehdaghi E, Banan M, Hosseinkhani S, Mohammadparast S, Oladnabi M, Ebrahimpour MR, Soosanabadi M, Farokhashtiani T, Darvish H, Firouzabadi SG, Farashi S, **Najmabadi H**, Ohadi M. Core promoter STRs: Novel mechanism for inter-individual variation in gene expression in humans. Gene. 2012; 492(1):195-8.

208. Neishabury M, Zamani S, Azarkeivan A, Abedini SS, Darvish H, Zamani F, **Najmabadi H**. The modifying effect of Xmn1-HBG2 on thalassemic phenotype is associated with its linked elements in the beta globin locus control region, including the palindromic site at 5'HS4. Blood Cells Mol Dis. 2012; 48(1):1-5.

209. Bazazzadegan N, Nikzat N, Fattahi Z, Nishimura C, Meyer N, Sahraian S, Jamali P, Babanejad M, Kashef A, Yazdan H, Sabbagh Kermani F, Taghdiri M, Azadeh B, Mojahedi F, Khoshaeen A, Habibi H, Reyhanifar F, Nouri N, Smith RJ, Kahrizi K, **Najmabadi H**. The spectrum of GJB2 mutations in the Iranian population with non-syndromic hearing loss-A twelve year study. Int J Pediatr Otorhinolaryngol. 2012; 76(8):1164-74.

210. Banan M, Bayat H, Azarkeivan A, Mohammadparast S, Kamali K, Farashi S, Bayat N, Khani MH, Neishabury M, **Najmabadi H**. The XmnI and BCL11A single nucleotide polymorphisms may help predict hydroxyurea response in Iranian β-Thalassemia patients. Hemoglobin. 2012; 36(4):371-80.

211. Abedini SS, Kahrizi K, Behjati F, Banihashemi S, Ghasemi Firoozabadi S, **Najmabadi H**. Mutational screening of ARX gene in Iranian families with X-linked intellectual disability. Arch Iran Med. 2012; 15 (6):361-5.

212. French VM, van de Laar IM, Wessels MW, Rohe C, Roos-Hesselink JW, Wang G, Frohn-Mulder IM, Severijnen LA, de Graaf BM, Schot R, Breedveld G, Mientjes E, van Tienhoven M, Jadot E, Jiang Z, Verkerk A, Swagemakers S, Venselaar H, Rahimi Z, **Najmabadi H**, Meijers-Heijboer H, de Graaff E, Helbing WA, Willemsen R, Devriendt K, Belmont JW, Oostra BA, Amack JD, Bertoli-Avella AM. NPHP4 variants are associated with pleiotropic heart malformations. Circ Res. 2012; 110(12):1564-74.

213. 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; 90(5):847-55.

214. Banan M, Esmaeilzadeh-Gharehdaghi E, Nezami M, Deilami Z, Farashi S, Philipsen S, Esteghamat F, Pourfarzad F, Ali Imam AM, **Najmabadi H**. CREB1 is required for hydroxyurea-mediated induction of γ-globin expression in K562 cells. Clin Exp Pharmacol Physiol. 2012; 39(6):510-7.

215. Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ,

Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, **Najmabadi H**, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizberea A, Soler DM, Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H. Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. J Neurol. 2012; 259(5):838-50.

216. Yazdanpanahi N, Chaleshtori MH, Tabatabaiefar MA, Noormohammadi Z, Farrokhi E, **Najmabadi H**, Shahbazi S, Hosseinipour A. Two novel SLC26A4 mutations in Iranian families with autosomal recessive hearing loss. Int J Pediatr Otorhinolaryngol. 2012; 76(6):845-50.

217. Khorshidi M, Roshan P, Bayat N, Mahdavi MR, **Najmabadi H**. Hemoglobin Q-Iran detected in family members from Northern Iran: a case report. J Med Case Rep. 2012; 6(1):47.

218. Davarnia B, Babanejad M, Fattahi Z, Nikzat N, Bazazzadegan N, Pirzade A, Farajollahi R, Nishimura C, Jalalvand K, Arzhangi S, Kahrizi K, Smith RJ, **Najmabadi H**. Int J Pediatr Otorhinolaryngol. 2012; 76(2):268-71.

219. Rafiq MA, Kuss AW, Puettmann 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; 89(1):176-82.

220. Nourouzi V, Azizi H, Esteghamat F, Bazazzadegan N, Fattahi Z, Babanejad M, Nishimura C, Nikzat N, Jalalvand Kh, Kahrizi K, Smith RJH, **Najmabadi H**. Did the GJB2 35delG mutation originates in Iran? Am J Med Genet A. 2011; 155A(10):2453-8.

221. Behjati F, Ghasemi Firouzabadi S, Kahrizi K, Kariminejad R, Bagherizadeh I, Ansari J, Fallah M, Mojtahedi F, Darvish H, Bahrami Monajemi G, Abedini SS, Jamali P, Mojahedi F, Zadeh-Vakili A, **Najmabadi H**. Chromosome abnormality rate among Iranian patients with idiopathic mental retardation from consanguineous marriages. Arch Med Sci. 2011; 7(2):321-5.

222. Esmaeilzadeh-Gharehdaghi E, Banan M, Farashi S, Mirabzadeh A, Farokhashtiani T, Hosseinkhani S, Heidari A, **Najmabadi H**, Ohadi M. Support for down-tuning of the calreticulin gene in the process of human evolution. Prog Neuropsychopharmacol Biol Psychiatry. 2011; 35(7):1770-3.

223. Heidari A, Behmanesh M, Sahraian MA, Meshkani R, Darvish H, **Najmabadi H**, Ohadi M. The human caveolin 1 gene upstream purine complex and neurodegeneration--a common signature. J Neuroimmunol. 2011; 236(1-2):106-10.

224. Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, **Najmabadi H**, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, Urtizberea A, von der Hagen M, Hübner A, Palace J, Bushby K, Straub V, Beeson D, Abicht A, Lochmüller H. Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. Am J Hum Genet. 2011; 88(2):162-72.

225. Daneshi A, Hassanzadeh S, Emamdjomeh H, Mohammadi SH, Arzhangi S, Farhadi M, **Najmabadi H**. Prevalence of GJB2-associated deafness and outcomes of cochlear implantation in Iran. J Laryngol Otol. 2011; 125(5):455-9.

226. Neishabury M, Azarkeivan A, Oberkanins C, Abedini SS, Zamani S, **Najmabadi H**. Analyzing 5'HS3 and 5'HS4 LCR core regions and NF-E2 in Iranian thalassemia intermedia patients with normal or carrier status for beta-globin mutations. Blood Cells Mol Dis. 2011; 46(3):201-5.

227. Darvish H, Nabi MO, Firouzabadi SG, Karimlou M, Heidari A, **Najmabadi H**, Ohadi M. Exceptional human core promoter nucleotide compositions. Gene. 2011; 475(2):79-86.

228. Borck G, Ur Rehman A, Lee K, Pogoda HM, Kakar N, von Ameln S, Grillet N, Hildebrand MS, Ahmed ZM,

Nürnberg G, Ansar M, Basit S, Javed Q, Morell RJ, Nasreen N, Shearer AE, Ahmad A, Kahrizi K, Shaikh RS, Ali RA, Khan SN, Goebel I, Meyer NC, Kimberling WJ, Webster JA, Stephan DA, Schiller MR, Bahlo M, **Najmabadi H**, Gillespie PG, Nürnberg P, Wollnik B, Riazuddin S, Smith RJ, Ahmad W, Müller U, Hammerschmidt M, Friedman TB, Riazuddin S, Leal SM, Ahmad J, Kubisch C. Loss-of-function mutations of ILDR1 cause autosomal-recessive hearing impairment DFNB42. Am J Hum Genet. 2011; 88(2):127-37.

229. Hu H, Eggers K, Chen W, Garshasbi M, Motazacker MM, Wrogemann K, Kahrizi K, Tzschach A, Hosseini M, Bahman I, Hucho T, Mühlenhoff M, Gerardy-Schahn R, **Najmabadi H**, Ropers HH, Kuss AW. ST3GAL3 mutations impair the development of higher cognitive functions. Am J Hum Genet. 2011; 89(3):407-14.

230. Bazazzadegan N, Sheffield AM, Sobhani M, Kahrizi K, Meyer NC, Van Camp G, Hilgert N, Abedini SS, Habibi F, Daneshi A, Nishimura C, Avenarius MR, Farhadi M, Smith RJ, **Najmabadi H**. Two iranian families with a novel mutation in GJB2 causing autosomal dominant nonsyndromic hearing loss. Am J Med Genet A. 2011; 155A(5):1202-11.

231. Schneider SA, Ramirez A, Shafiee K, Kaiser FJ, Erogullari A, Brüggemann N, Winkler S, Bahman I, Osmanovic A, Shafa MA, Bhatia KP, **Najmabadi H**, Klein C, Lohmann K. Homozygous THAP1 mutations as cause of early-onset generalized dystonia. Mov Disord. 2011; 26(5):858-61.

232. Borck G, Ur Rehman A, Lee K, Pogoda HM, Kakar N, von Ameln S, Grillet N, Hildebrand MS, Ahmed ZM, Nürnberg G, Ansar M, Basit S, Javed Q, Morell RJ, Nasreen N, Shearer AE, Ahmad A, Kahrizi K, Shaikh RS, Ali RA, Khan SN, Goebel I, Meyer NC, Kimberling WJ, Webster JA, Stephan DA, Schiller MR, Bahlo M, **Najmabadi H**, Gillespie PG, Nürnberg P, Wollnik B, Riazuddin S, Smith RJ, Ahmad W, Müller U, Hammerschmidt M, Friedman TB, Riazuddin S, Leal SM, Ahmad J, Kubisch C. Loss-of-function mutations of ILDR1 cause autosomal-recessive hearing impairment DFNB42. Am J Hum Genet. 2011; 88(2):127-37.

233. Smith KR, Bromhead CJ, Hildebrand MS, Shearer AE, Lockhart PJ, **Najmabadi H**, Leventer RJ, McGillivray G, Amor DJ, Smith RJ, Bahlo M. Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biol. 2011; 12(9):R85.

234. Rafiq MA, Kuss AW, Puettmann 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; 89(1):176-82.

235. 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; 155A(8):1976-80.

236. 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; 19(1):115-7.

237. Kuss AW, Garshasbi M, Kahrizi K, Tzschach A, Behjati F, Darvish H, Abbasi-Moheb L, Puettmann L, Zecha A, Weissmann R, Hu H, Mohseni M, Abedini SS, Rajab A, Hertzberg C, Wieczorek D, Ullmann R, Ghasemi-Firouzabadi S, Banihashemi S, Arzhangi 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; 129(2):141-8.

238. Ebrahimkhani S, Azarkeivan A, Bayat N, Houry-Parvin M, Jalil-Nejad S, Zand S, Golkar Z, Hadavi V, Imanian H, Oberkanins C, **Najmabadi H**. Genotype-phenotype correlation in Iranian patients with hemoglobin H disease. Hemoglobin. 2011; 35(1):40-6.

239. Farokhashtiani T, Mirabzadeh A, Olad Nabi M, Magham ZG, Khorshid HR, **Najmabadi H**, Ohadi M. Reversion of

the human calreticulin gene promoter to the ancestral type as a result of a novel psychosis-associated mutation. Prog Neuropsychopharmacol Biol Psychiatry. 2011; 35(2):541-4.

240. 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 H-H, **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. PNAS. 2011; 108:12390-5.

241. **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 MJS, 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; 478:57-63.

242. Hildebrand MS, Kahrizi K, Bromhead CJ, Shearer AE, Webster JA, Khodaei H, Abtahi R, Bazazzadegan N, Babanejad M, Nikzat N, Kimberling WJ, Stephan D, Huygen PL, Bahlo M, Smith RJ, **Najmabadi H**. Mutations in TMC1 are a common cause of DFNB7/11 hearing loss in the Iranian population. Ann Otol Rhinol Laryngol. 2010; 119(12):830-5.

243. de Souza Rocha Simonini P, Breiling A, Gupta N, Malekpour M, Youns M, Omranipour R, Malekpour F, Volinia S, Croce CM, **Najmabadi H**, Diederichs S, Sahin O, Mayer D, Lyko F, Hoheisel JD, Riazalhosseini Y. Epigenetically deregulated microRNA-375 is involved in a positive feedback loop with estrogen receptor alpha in breast cancer cells. Cancer Res. 2010; 70(22):9175-84.

244. Bidari A, Ghavidel-Parsa B, **Najmabadi H**, Talachian E, Haghighat-Shoar M, Broumand B, Ghalehbaghi B. Common MEFV mutation analysis in 36 Iranian patients with familial Mediterranean fever: clinical and demographic significance. Mod Rheumatol. 2010; 20(6):566-72.

245. de Souza Rocha Simonini P, Breiling A, Gupta N, Malekpour M, Youns M, Omranipour R, Malekpour F, Volinia S, Croce CM, **Najmabadi H**, Diederichs S, Sahin O, Mayer D, Lyko F, Hoheisel JD, Riazalhosseini Y. Epigentically deregulated microRNA-375 is involved in a positive feedback loop with Estrogen Receptor α in breast cancer cells. Cancer Res. 2010; 70(22):9175-84.

246. Yang T, Kahrizi K, Bazazzadeghan N, Meyer N, **Najmabadi H**, Smith RJ. A novel mutation adjacent to the Bth mouse mutation in the TMC1 gene makes this mouse an excellent model of human deafness at the DFNA36 locus. Clin Genet. 2010; 77(4):395-8.

247. Shafeghati Y, Kahrizi K, **Najmabadi H**, Kuss AW, Ropers HH, Tzschach A. Brachyphalangy, polydactyly and tibial aplasia/hypoplasia syndrome (OMIM 609945): case report and review of the literature. Eur J Pediatr. 2010; 169(12):1535-9.

248. Darvish H, Esmaeeli-Nieh S, Monajemi GB, Mohseni M, Ghasemi-Firouzabadi S, Abedini SS, Bahman I, Jamali P, Azimi S, Mojahedi F, Dehghan A, Shafeghati Y, Jankhah A, Falah M, Soltani Banavandi MJ, Ghani-Kakhi M, Garshasbi M, Rakhshani F, Naghavi A, Tzschach A, Neitzel H, Ropers HH, Kuss AW, Behjati F, Kahrizi K, **Najmabadi H**. A Clinical and Molecular Genetic study of 112 Iranian families with primary microcephaly. J Med Genet. 2010; 47(12):823-8.

249. Azarkeivan A, Neishabury M, Hadavi V, Esteghamat F, Enrahimkhani S, **Najmabadi H**. A report of 8 cases with hemoglobin H disease in an Iranian family. Pediatr Hematol Oncol. 2010; 27(5):405-12.

250. Jalal SD, Al-Allawi NAS, **Najmabadi H**, Bayat N, Imanian H, Faraj A. β-thalassemia mutation in the Kurdish population of Northeastern Iraq. J Hemoglobin. 2010; 34(5): 469-76.

251. Hildebrand MS, Witmer PD, Xu S, Newton SS, Kahrizi K, **Najmabadi H**, Valle D, Smith RJ. miRNA mutations are

not a common cause of deafness. Am J Med Genet A. 2010; 152A(3):646-52.

252. Odeh H, Hunker KL, Belyantseva IA, Azaiez H, Avenarius MR, Zheng L, Peters LM, Gagnon LH, Hagiwara N, Skynner MJ, Brilliant MH, Allen ND, Riazuddin S, Johnson KR, Raphael Y, **Najmabadi H**, Friedman TB, Bartles JR, Smith RJ, Kohrman DC. Mutations in Grxcr1 are the basis for inner ear dysfunction in the pirouette mouse. Am J Hum Genet. 2010; 86(2):148-60.

253. Zarif Yeganeh M, Mirabzadeh A, Khorram Khorshid HR, Kamali K, Heshmati Y, Gozalpour E, Veissy K, Olad Nabi M, **Najmabadi H**, Ohadi M. Novel extreme homozygote haplotypes at the human caveolin 1 gene upstream purine complex in sporadic Alzheimer's disease. Am J Med Genet B Neuropsychiatr Genet. 2010; 153B(1):347-9.

254. Hildebrand MS, Thorne NP, Bromhead CJ, Kahrizi K, Webster JA, Fattahi Z, Bataejad M, Kimberling WJ, Stephan D, **Najmabadi H**, Bahlo M, Smith RJ. Variable hearing impairment in a DFNB2 family with a novel MYO7A missense mutation. Clin Genet. 2010; 77(6):563-71.

255. Hildebrand MS, Avenarius MR, Fellous M, Zhang Y, Meyer NC, Auer J, Serres C, Kahrizi K, **Najmabadi H**, Beckmann JS, Smith RJ. Genetic male infertility and mutation of CATSPER ion channels. Eur J Hum Genet. 2010; 18(11):1178-84.

256. Nabi MO, Mirabzadeh A, Feizzadeh G, Khorshid HR, Karimlou M, Yeganeh MZ, Asgharian AM, **Najmabadi H**, Ohadi M. Novel mutations in the calreticulin gene core promoter and coding sequence in schizoaffective disorder. Am J Med Genet B Neuropsychiatr Genet. 2010; 153B(2):706-9.

257. Neishabury M, Azarkeivan A, **Najmabadi H**. Frequency of positive XmnIGgamma polymorphism and coinheritance of common alpha thalassemia mutations do not show statistically significant difference between thalassemia major and intermedia cases with homozygous IVSII-1 mutation. Blood Cells Mol Dis. 2010; 44(2):95-9.

258. Hasanzad M, Azad M, Kahrizi K, Saffar BS, Nafisi S, Keyhanidoust Z, Azimian M, Refah AA, Also E, Urtizberea JA, Tizzano EF, **Najmabadi H**. Carrier frequency of SMA by quantitative analysis of the SMN1 deletion in the Iranian population. Eur J Neurol. 2010; 17(1):160-2.

259. Mir A, Kaufman L, Noor A, Motazacker MM, Jamil T, Azam M, Kahrizi K, Rafiq MA, Weksberg R, Nasr T, Naeem F, Tzschach A, Kuss AW, Ishak GE, Doherty D, Ropers HH, Barkovich AJ, **Najmabadi H**, Ayub M, Vincent JB. Identification of mutations in TRAPPC9, which encodes the NIK- and IKK-beta-binding protein, in nonsyndromic autosomal-recessive mental retardation. Am J Hum Genet. 2009; 85(6):909-15.

260. Zarif Yeganeh M, Ghaffarpour M, Farhud DD, Karimlou M, Ghabaee M, Haghighi Nazari A, **Najmabadi H**, Ohadi M. Skew in the human caveolin 1 gene upstream purine complex homozygote haplotype compartment in multiple sclerosis. J Neuroimmunol. 2009; 216(1-2):103-7.

261. Grillet N, Schwander M, Hildebrand MS, Sczaniecka A, Kolatkar A, Velasco J, Webster JA, Kahrizi K, **Najmabadi H**, Kimberling WJ, Stephan D, Bahlo M, Wiltshire T, Tarantino LM, Kuhn P, Smith RJ, Müller U. Mutations in LOXHD1, an evolutionarily conserved stereociliary protein, disrupt hair cell function in mice and cause progressive hearing loss in humans. Am J Hum Genet. 2009; 85(3):328-37.

262. Hadavi V, Jafroodi M, Hafezi-Nejad N, Moghadam SD, Eskandari F, Tarashohi S, Pourfahim H, Oberkanins C, Law HY, **Najmabadi H**. Alpha-thalassemia mutations in Gilan Province, North Iran. Hemoglobin. 2009; 33(3):235-41.

263. Avenarius MR, Hildebrand MS, Zhang Y, Meyer NC, Smith LL, Kahrizi K, **Najmabadi H**, Smith RJ. Human male infertility caused by mutations in the CATSPER1 channel protein. Am J Hum Genet. 2009; 84(4):505-10.

264. Tamaddoni A, Hadavi V, Nejad NH, Khosh-Ain A, Siami R, Aghai-Meibodi J, Almadani N, Oberkanins C, Law HY, **Najmabadi H**. alpha-Thalassemia mutation analyses in Mazandaran province, North Iran. Hemoglobin. 2009;

33(2):115-23.

265. Pouya AR, Abedini SS, Mansoorian N, Behjati F, Nikzat N, Mohseni M, Nieh SE, Abbasi L, Darvish H, Monajemi GB, Banihashemi S, Kahrizi K, Ropers HH, **Najmabadi H**. Fragile X Syndrome Screening of Families with Consanguineous and Non-consanguineous Parents in the Iranian Population. Eur J Med Genet. 2009; 52(4):170-3.

266. Shearer AE, Hildebrand MS, Webster JA, Kahrizi K, Meyer NC, Jalalvand K, Arzhanginy S, Kimberling WJ, Stephan D, Bahlo M, Smith RJ, **Najmabadi H**. Mutations in the first MyTH4 domain of MYO15A are common cause of DFNB3 hearing loss. Laryngoscope. 2009; 119(4):727-33.

267. Hasanzad M, Golkar Z, Kariminejad R, Hadavi V, Almadani N, Afroozan F, Salahshurifar I, Shafeghati Y, Kahrizi K, **Najmabadi H**. Deletions in the survival motor neuron gene in Iranian patients with spinal muscular atrophy. Ann Acad Med Singapore. 2009; 38(2):139-41.

268. Shearer AE, Hildebrand MS, Bromhead CJ, Kahrizi K, Webster JA, Azadeh B, Kimberling WJ, Anousheh A, Nazeri A, Stephan D, **Najmabadi H**, Smith RJ, Bahlo M. A novel splice site mutation in the RDX gene causes DFNB24 hearing loss in an Iranian family. Am J Med Genet A. 2009; 149A(3):555-8.

269. Al-Allawi NA, Badi AI, Imanian H, Nikzat N, Jubrael JM, **Najmabadi H**. Molecular characterization of alpha-thalassemia in the Dohuk region of Iraq. Hemoglobin. 2009; 33(1):37-44.

270. 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, Christian Hennies H, Horn D. Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants of COH1. Hum Mutat. 2009; 30(2):E404-20.

271. 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; 17(1):125-8.

272. Kahrizi K, Mohseni M, Nishimura C, Bazazzadegan N, Fischer SM, Dehghani A, Sayfati M, Taghdiri M, Jamali P, Smith RJ, Azizi F, **Najmabadi H**. Identification of SLC26A4 gene mutations in Iranian families with hereditary hearing impairment. Eur J Pediatr. 2009; 168(6):651-3.

273. Heshmati Y, Mirabzadeh A, Feizzade G, Gilanipour M, Etminan MR, Khoram Khorshid HR, Kamali K, Fakhri M, Moghimi N, **Najmabadi H**, Ohadi M. A novel polymorphic purine complex at the 1.5 kb upstream region of the human caveolin-1 gene and risk of Alzheimer's disease; Extra-short alleles and accumulated allele homozygosity. Am J Med Genet B Neuropsychiatr Genet. 2009; 150B(2):248-53.

274. Hilgert N, Kahrizi K, Dieltjens N, Bazazzadegan N, **Najmabadi H**, Smith RJ, Van Camp G. A large deletion in GPR98 causes type IIC Usher syndrome in male and female members of an Iranian family. J Med Genet. 2009; 46(4):272-6.

275. Shearer AE, Hildebrand MS, Bromhead CJ, Kahrizi K, Webster JA, Azadeh B, Kimberling WJ, Anousheh A, Nazeri A, Stephan D, **Najmabadi H**, Smith RJ, Bahlo M. A novel splice site mutation in the RDX gene causes DFNB24 hearing loss in an Iranian family. Am J Med Genet A. 2009; 149A(3):555-8.

276. Turkmen S, Guo G, Garshasbi M, Hoffmann K, Kahrizi K, Shallah AA, Kuss A, Ropers H, Humphrey N, **Najmabadi H**, Mundlos S, Robinson P. Mutation of CA8 cause a novel syndrome characterized by ataxia and mild mental retardation with predisposition to quadrupedal gait. Accepted PLOS genetics, 2009.

277. Zhang Y, Malekpour M, Al-Madani N, Kahrizi K, Zanganeh M, Mohseni M, Mojahedi F, Daneshi A, **Najmabadi H**, Smith RJ. Sensorineural deafness and male infertility: a contiguous gene deletion syndrome. BMJ Case Rep. 2009; bcr08.2008.0645.

278. Hasanzad M, Golkar Z, Hadavi V, Kariminejad R, Almadani N, Afroozan F, Salahshurifar I, Shafeghati Y, Kahrizi K, **Najmabadi H**. Survival Motor Neuron Gene Deletions among Iranian Spinal Muscular Atrophy. Ann Acad Med Singapore. 2009; 38(2):139-41.

279. Neishabury M, Azarkeivan A, Oberkanins C, Esteghamat F, Amirizadeh N, **Najmabadi H**. Molecular mechanisms underlying thalassemia intermedia in Iran. Genet Test. 2008; 12(4):549-56.

280. Du X, Schwander M, Moresco EM, Viviani P, Haller C, Hildebrand MS, Pak K, Tarantino L, Roberts A, Richardson H, Koob G, **Najmabadi H**, Ryan AF, Smith RJ, Müller U, Beutler B. A catechol-O-methyltransferase that is essential for auditory function in mice and humans. Proc Natl Acad Sci U S A. 2008; 105(38):14609-14.

281. Zandian K, Nateghi J, Keikhaie B, Pedram M, Hafezi-Nejad N, Hadavi V, Oberkanins C, Azarkeivan A, Law HY, **Najmabadi H**. Alpha-thalassemia Mutations in Khuzestan Province, southwest Iran. Hemoglobin. 2008; 32(6):546-52.

282. Nikuei P, Hadavi V, Rajaei M, Saberi M, Hajizade F, **Najmabadi H**. Prenatal Diagnosis for-beta-thalassemia Major in the Iranian Province of Hormozgan. Hemoglobin 2008; 32(6):539-45.

283. Behjati F, Shafeghati Y, Kahrizi K, Firouzabadi SG, **Najmabadi H**, Dixon N, Davies AF. Interstitial deletion of the short arm of chromosome 10 del(10)(p11.2p12.32) in a patient with congenital heart disease, minor dysmorphism, and mental retardation. Am J Med Genet A. 2008; 146A(24):3223-6.

284. Khatami Sh, Rouhi Dehboneh S, Sadeghi S, Saeidi P, Mirzazadeh R, Bayat P, Amirkhani A, Samavat A, Zeinali S, Akbari MT, **Najmabadi H**. Globin chain synthesis for differential diagnosis of β-thalassemia from α-thalassemia carriers. Blood 2008; 4(4):239-46.

285. Nunes A, Ohadi M, Rahimi A, Aghajani A, **Najmabadi H**, Currais A, Soriano S. A mutation in the calreticulin gene promoter in a family case of schizoaffective disorder leads to its aberrant transcriptional activation. Brain Res. 2008; 1239:36-41.

286. Du X, Schwander M, Moresco EM, Viviani P, Haller C, Hildebrand MS, Pak K, Tarantino L, Roberts A, Richardson H, Koob G, **Najmabadi H**, Ryan AF, Smith RJ, Müller U, Beutler B. A catechol-O-methyltransferase that is essential for auditory function in mice and humans. Proc Natl Acad Sci U S A 2008; 105(38):14609-14.

287. Naghavi A, Nishimura C, Kahrizi K, Riazalhosseini Y, Bazazzadegan N, Mohseni M, Smith RJ, **Najmabadi H**. GJB2 mutations in Baluchi population. J Genet. 2008; 87(2):195-7.

288. Behjati F, Shafaghati Y, Firouzabadi SG, Kahrizi K, Bagherizadeh I, **Najmabadi H**, Bint S, Ogilvie C. M-banding characterization of a 16p11.2p13.1 tandem duplication in a child with autism, neurodevelopmental delay and

dysmorphism. Eur J Med Genet. 2008; 51(6):608-14.

289. Tzschach A, Bozorgmehr B, Hadavi V, Kahrizi K, Garshasbi M, Motazacker MM, Ropers HH, Kuss AW, **Najmabadi H**. Alopecia-mental retardation syndrome: clinical and molecular characterization of four patients. Br J Dermatol. 2008; 159(3):748-51.

290. Shojasaffar B, Moradin N, Kahrizi K, Cobo AM, **Najmabadi H**. CTG expansion & haplotype analysis in DM1 gene in healthy Iranian population. Can J Neurol Sci. 2008; 35(2):216-9.

291. Garshasbi M, Hadavi V, Habibi H, Kahrizi K, Kariminejad R, Behjati F, Tzschach A, **Najmabadi H**, Ropers HH, Kuss AW. A Defect in the TUSC3 Gene is Associated with Autosomal Recessive Mental Retardation. Am J Hum Genet. 2008; 82(5):1158-64.

292. Banoei MM, Majidizadeh T, Shirazi E, Moghimi N, Ghadiri M, **Najmabadi H**, Ohadi M. No association between the DAT1 10-repeat allele and ADHD in the Iranian population. Am J Med Genet B Neuropsychiatr Genet. 2008; 147(1):110-1.

293. Moheb LA, Tzschach A, Garshasbi M, Kahrizi K, Darvish H, Heshmati Y, Kordi A, **Najmabadi H**, Ropers HH, Kuss AW. Identification of a nonsense mutation in the very low-density lipoprotein receptor gene (VLDLR) in an Iranian family with dysequilibrium syndrome. Eur J Hum Genet. 2008; 16(2):270-3.

294. **Najmabadi H**, Motazacker MM, Garshasbi M, Kahrizi K, Tzschach A, Chen W, Behjati F, Hadavi V, Nieh SE, Abedini SS, Vazifehmand R, Firouzabadi SG, Jamali P, Falah M, Seifati SM, Grüters A, Lenzner S, Jensen LR, Rüschendorf F, Kuss AW, Ropers HH. Homozygosity mapping in consanguineous families reveals extreme heterogeneity of non-syndromic autosomal recessive mental retardation and identifies 8 novel gene loci. Hum Genet. 2007; 121:143-8.

295. Mani A, Radhakrishnan J, Wang H, Mani A, Mani M-A, Nelson-Williams C, Carew KS, Mane S, **Najmabadi H**, Wu D, Lifton RP. LRP6 mutation in a family with early coronary disease and metabolic risk factors. Science. 2007; 315:1278-82.

296. Abolghasemi H, Amid A, Zeinali S, Radfar MH, Eshghi P, Rahiminejad MS, Ehsani MA, **Najmabadi H**, Akbari MT, Afrasiabi A, Akhavan-Niaki H, Hoorfar H. Thalassemia in Iran: Epidemiology, prevention, and management. J Pediatr Hematol Oncol. 2007; 29(4):233-8.

297. Ohadi M, Keikhaee MR, Javanbakht A, Sargolzaee MR, Robabeh M, **Najmabadi H**. Gender dimorphism in the DAT1 -67 T-allele homozygosity and predisposition to bipolar disorder. Brain Res. 2007; 1144:142-5.

298. Esteghamat F, Imanian H, Azarkeivan A, Banan M, Pourfarzad F, Almadani N, **Najmabadi H**. Iranian δβ-Thalassemia Patients for the eight most deletions. Hemoglobin. 2007; 31(4):463-9.

299. Salahshourifar I, Shafeghati Y, Golkar Z, **Najmabadi H**. Molecular analysis of the neuronal apoptosis inhibitory protein gene in families with spinal muscular atrophy. Arch Iran Med. 2007; 10(4):509-13.

300. Motazacker MM, Rost BR, Hucho T, Garshasbi M, Kahrizi K, Ullmann R, Abedini SS, Nieh SE, Amini SH, Goswami C, Tzschach A, Jensen LR, Schmitz D, Ropers HH, **Najmabadi H**, Kuss AW. A Defect in the ionotropic glutamate receptor 6 gene (GRIK2) is associated with autosomal recessive mental retardation. Am J Hum Genet. 2007; 81(4):792-8.

301. Khatami S, Dehboneh SR, Sadeghi S, Mirzazadeh R, Saeedi P, Bayat P, **Najmabadi H**, Zeinali S, Akbari MT,

Ardjmand M, Amirkhani A. Globin Chain Synthesis is a Useful Complementary Tool in the Differential Diagnosis of Thalassemias. Hemoglobin. 2007; 31(3):333-41.

302. Hadavi V, Taromchi AH, Malekpour M, Gholami B, Law HY, Almadani N, Afroozan F, Sahebjam F, Pajouh P, Kariminejad R, Kariminejad MH, Azarkeivan A, Jafroodi M, Tamaddoni A, Puehringer H, Oberkanins C, **Najmabadi H**. Elucidating the spectrum of alpha-thalassemia mutations in Iran. Haematologica. 2007; 92(7):992-3.

303. Motazacker MM, Taherzadeh-Fard E, Husseini Z, Behjati F, Esteghamat F, Kahrizi K, **Najmabadi H**. The Iranian human mutation database. Arch Iran Med. 2007; 10(3):372-5.

304. Meyer NC, Alasti F, Nishimura CJ, Imanirad P, Kahrizi K, Riazalhosseini Y, Malekpour M, Kochakian N, Jamali P, Van Camp G, Smith RJ, **Najmabadi H**. Identification of three novel TECTA mutations in Iranian families with autosomal recessive nonsyndromic hearing impairment at the DFNB21 locus. Am J Med Genet A. 2007; 143(14):1623-9.

305. Van Laer L, Meyer NC, Malekpour M, Riazalhosseini Y, Moghannibashi M, Kahrizi K, Vandevelde A, Alasti F, **Najmabadi H**, Van Camp G, Smith RJ. A novel DFNA5 mutation does not cause hearing loss in an Iranian family. J Hum Genet. 2007; 52(6):549-52.

306. Schwander M, Sczaniecka A, Grillet N, Bailey JS, Avenarius M, **Najmabadi H**, Steffy BM, Federe GC, Lagler EA, Banan R, Hice R, Grabowski-Boase L, Keithley EM, Ryan AF, Housley GD, Wiltshire T, Smith RJ, Tarantino LM, Muller U. A forward genetics screen in mice identifies recessive deafness traits and reveals that pejvakin is essential for outer hair cell function. J Neurosci. 2007; 27(9):2163-75.

307. Rezaie T, Karimi-Nejad MH, Meshkat MR, Sohbati S, Karimi-Nejad R, **Najmabadi H**, Sarfarazi M. Genetic screening of leber congenital amaurosis in a large consanguineous Iranian family. Ophthalmic Genet. 2007; 28(4):224-8.

308. Puehringer H, **Najmabadi H**, Law HY, Krugluger W, Viprakasit V, Pissard S, Baysal E, Taher A, Farra Ch, Al-Ali A, Al-Ateeq S, Obercanins Ch. Validation of reverse-hybridization StripAssay for the simultaneous analysis of common α-thalassemia point mutations and deletions. Clin Chem Lab Med. 2007; 45(5):605-10.

309. Ohadi M, Keikhaee MR, Javanbakht A, Sargolzaee MR, Mazinani R, **Najmabadi H**. Gender dismorphism in the DAT1-67 T-allele homozygosity and predisposition to biopolar disorder. Brain Res. 2007; 1144:142-5.

310. Wilson RC, Nimkarn S, Dumic M, Obeid J, Azar M, **Najmabadi H**, Saffari F, New MI. Ethnic-specific distribution of mutations in 716 patients with congenital adrenal hyperplasia owing to 21-hydroxylase deficiency. Mol Genet Metab. 2007; 90(4):414-21.

311. Ohadi M, Totonchi M, Maguire P, Lindblom A, Habibi R, Afshin Alavi B, Keyhani E, **Najmabadi H**. Mutation analysis of the DBC2 gene in sporadic and familial breast cancer. Acta Oncol. 2007; 46(6):770-2.

312. Zhang Y, Malekpour M, Al-Madani N, Kahrizi K, Zanganeh M, Mohseni M, Mojahedi F, Daneshi A, **Najmabadi H**, Smith RJ. Sensorineural deafness and male infertility-a contiguous gene deletion syndrome. J Med Genet. 2007; 44(4):233-40.

313. Keikhaee MR, Hashemi SB, **Najmabadi H**, Noroozian M. C677T methylentetrahydrofulate reductase and angiotensin converting enzyme gene polymorphisms in patients with Alzheimer's disease in Iranian population. Neurochem Res. 2006; 31(8):1079-83.

314. Ohadi M, Shirazi E, Tehranidoosti M, Moghimi N, Keikhaee MR, Ehssani S, Aghajani A, **Najmabadi H**. Attention-deficit/hyperactivity disorder (ADHD) association with the DAT1 core promoter -67 T allele. Brain Res. 2006; 1101(1):1-4.

315. Excoffon KJDA, Avenarius MR, Hansen MR, Kimberling WJ, **Najmabadi H**, Smith RJH. Detection and

Characterization of Mutations in the Coxsackievirus and Adenovirus Receptor (CAR). Hear Res. 2006; 215(1-2):1-9.

316. Kleanthous M, Patsalis PC, Drousiotou A, Motazacker M, Christodoulou K, Cariolou M, Baysal E, Khrizi K, Moghimi B, Pourfarzad F, van Baal S, Deltas C, **Najmabadi H**, Patrinos GP. The Cypriot and Iranian National Mutation Frequency Databases Online Citation: Human Mutation. 2006; 27(6):598-9.

317. Bajestan SN, Sabouri AH, Nakamura M, Takashima H, Keikhaee MR, Behdani F, Fayyazi MR, Sargolzaee MR, Bajestan MN, Sabouri Z, Khayami E, Haghighi S, Hashemi SB, Eiraku N, Tufani H, **Najmabadi H**, Arimura K, Sano A, Osame M. Association of AKT1 haplotype with the risk of schizophrenia in Iranian population. Am J Med Genet B Neuropsychiatr Genet. 2006; 141(4):383-6.

318. Aghajani A, Rahimi A, Fadai F, Ebrahimi A, **Najmabadi H**, Ohadi M. A point mutation at the calreticulin gene core promoter conserved sequence in a case of schizophrenia. Am J Med Genet B Neuropsychiatr Genet. 2006; 141(3):294-5.

319. Law HY, Luo HY, Wang W, Ho JF, **Najmabadi H**, Ng IS, Steinberg MH, Chui DH, Chong SS. Determining the cause of patchwork HBA1 and HBA2 genes: recurrent gene conversion or crossing over fixation events. Haematologica. 2006; 91(3):297-302.

320. **Najmabadi H**, Ghamari A, Sahebjam F, Kariminejad R, Hadavi V, Khatibi T, Samavat A, Mehdipour E, Modell B, Kariminejad MH. Fourteen-Year Experience of Prental Diagnosis of Thalassemia in Iran. Community Genet. 2006; 9(2):93-7.

321. **Najmabadi H**, Nishimura C, Kahrizi K, Riazalhosseini Y, Malekpour M, Daneshi A, Farhadi M, Mohseni M, Mahdieh N, Ebrahimi A, Bazazzadegan N, Naghavi A, Avenarius M, Arzhangi S, Smith RJH. GJB2 mutations: Passage through Iran. Am J Med Genet. 2005; 133 A:132-7.

322. Behjati F, Atri M, **Najmabadi H**, Nouri K, Zamani M, Mehdipour P. Prognostic Value of Chromosome 1 and 8 Copy Number in Invasive Ductal Breast Carcinoma Among Iranian Women: An Interphase FISH Analysis. Pathol Oncol Res. 2005;11(3):157-63.

323. Keikhaee MR, Fadai F, Sargolzaee MR, Javanbakht A, **Najmabadi H**, Ohadi M. Association analysis of the dopamine Transporter (DAT1)-67A/T polymorphism in bipolar disorder. Am J Med Genet PartB (Neuropsychiatric Genetics) 2005; 135B(1):47-9.

324. Riazalhosseini Y, Nishimura C, Kahrizi K, Shafeghati Y, Joghataie MT, Mohseni M, Mahdieh N, Javan MK, Smith RH, **Najmabadi H**. DELTA (GJB6-D3S1830) is not common cause of nonsyndromic Hearing loss in the Iranian Population. Arch in Irn Med. 2005; 8(2):104–8.

325. Shafeghati Y, Teimourian Sh, Babamohammadi Gh, Afrouzan F, Almadani SN, Karimi-Nejad R, **Najmabadi H**. Association between the DRD2 A1 Allele and opium-Addiction in the Iranian population. Am J Med Genet B Neuropsychiatr Genet. 2005; 134(1):39-41.

326. Garshasbi M, Motazacker MM, Kahrizi K, Behjati F, Abedini SS, Esmaeeli Nieh S, Firouzabadi SG, Becker C, Rueschendorf F, Nurnberg P, Tzschach A, Vazifemand R, Erdogan F, Ullmann R, Lenzner S, Kuss AW, Ropers HH, **Najmabadi H**. SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. Hum Genet. 2006; 118(6):708-15.

327. Chen W, Kahrizi K, Meyer NC, Riazalhosseini Y, Mueller RF, McGuirt WT, Van Camp G, **Najmabadi H**, JH Smith R. Mutation of COL11A2 causes Autosomal Ressesive non-syndromic hearing loss at DFNB53 Locus. J Med Genet. 2005; 42(10):e61.

328. Kahrizi K, Nishimura C, Naghavi A, Riazalhosseini Y, JH Smith R, **Najmabadi H**. A novel mutation of SLC26A4 gene in an Iranian family with Pendred syndrome. Int J Endocrinol Metab. 2005; 2:104-8.

329. Radpour R, Haghighi MM, Ohadi M, Broumand B, **Najmabadi H**. Molecular Study of PKD1 & PKD2 Genes by Linkage Analysis and Determining the Genotype/Phenotype Correlations in Several Iranian Families with Autosomal Dominant Polycystic Kidney. Med J Islam Rep Iran. 2005; 19(1):65-75.

330. Moghimi B, Yavari M, Oberkanins Ch, Hosseini Amini SS, Khatami S, Rouhi S, Kahrizi K, **Najmabadi H**. Hb DHONBURI (Neapolis) [β 126 (H4) Val→ Gly] Identified in A Family From Northern Iran. Haemoglobin. 2004; 28(4):353-6.

331. Shafeghati Y, Teimourian Sh, Babamohammadi Gh, Afrouzan F, Almadani SN, Karimi-Nejad R, **Najmabadi H**. Molecular Diagnosis in Iranian Patients with Spinal Muscular Atrophy. Arch Irn Med. 2004; 7(1): 47-52.

332. 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. 2004; 129B(1):10-12.

333. Mahdieh N, Nichimura C, Ali Madadi K, Riazalhosseini Y, Yazdan H, Arzhangi S, Jalalvand K, Ebrahimi A, Kazemi S, JH Smith R, **Najmabadi H**. The Frequency of GJB2 mutations and the Δ(GJB6-D13S1830) Deletion as A Cause of Autosomal Recessive Non-Syndromic Deafness in the Kurdish Population. Clin Genet. 2004; 65(6): 106-508.

334. **Najmabadi H,** Neishabury M, Sahebjam F, Kahrizi K, Shafaghati Y, Nikzat N, Jalalvand M, Aminy F, Hashemi SB, Moghimi B, Reza Noorian A, Jannati A, Mohammadi M, Javan K. The Iranian Human Mutation Gene Bank: A data and sample resource for worldwide collaborative genetics research. Hum Mut. 2003; 21:146-50.

335. Krause S, Schlottel-Weigel B, Walter C, **Najmabadi H**, Wiendl H, Müller-Höcker J, Muller-Felber W, Pongratz DE, Lochmuller H. A novel homozygous missense mutation in the GNE Gene of a patient with quadriceps spring recessive hereditary inclusion body Myopathy associated with muscle inflammation. Neuromuscul Disord. 2003; 13(10):830-4.

336. Garshasbi M, Oberkanins Ch, Law HY, Neishabury M, Karimi-Nejad R, **Najmabadi H**. α-Globin Gene Deletion and Point mutation Analysis Among Iranian Patients with Microceytic Hypochromic Anemia. J Hematol. 2003; 88(10):1196-7.

337. Neishaboury M, Oberkanins Ch, Abbasi Moheb L, Pourfathollah AA, Kahrizi K, Keyhani E, Krugluger W, **Najmabadi H**. High prevalence of the -α3.7 Deletion Among Thalassemia Patients in Iran. Hemoglobin. 2003; 27(1):53-5.

338. **Najmabadi H**, Poorfathollah AA, Neishabury M, Sahebjam F, Krugluger W, Oberkanins Ch. Rare and Unexpected Mutations Among Iranian Beta-Thalassemia patient and Prenatal Samples discovery by Reverse-Hybridization and Sequencing. Haematologica. 2002; 87(10):1113-4.

339. Pasalar P, **Najmabadi H**, Noorian A, Moghimi B, Jannati A, Soltanzadeh A, Krefft T, Crook R, Hardi J. A family with Alzheimer's disease caused by a Novel APP Mutation (THR714ALA). Neurology. 2002; 58(10):1574-5.

340. **Najmabadi H**, Cucci R, Sahebjam S, Kouchakian N, Farhadi M, Kahrizi K, Arzhangi S, Daneshmandan N, Kh Javan M, JH Smith R. GJB2 mutations in Iranians with autosomal recessive non-syndromic sensorineural hearing loss. Hum Mutat. 2002; 19(5):572.

341. Neishabury M, Abbasi Moheb L, Poorfathollah AA, Kahrizi K, Keyhany E, Krugluger W, Obercanins Ch, **Najmabadi H**. Alpha Thalassemia Deletion Analysis in Iran. Arch Irn Med. 2001; 4(4):160-4.

342. Karimi Nejad MH, Meshkat MR, Sohbati S, Rezaei T, Karimi Nejad R, **Najmabadi H**, Sarfarazi M. Report of Leber Congenital Amaurosis in a Large Iranian Kindred. Arch Irn Med. 2001; 4(4):171-6.

343. **Najmabadi H**, et al. Detection of Yq Microdeletion in oligozoospermic men. Med J Islam Rep Iran. 2001; 14:119-25.

344. **Najmabadi H**, Karimi-Nejad R, Sahebjam S, Pourfarzad F, Teimourian S, Sahebjam F, Amirizadeh N, Karimi-Nejad MH. The β Thalasemia Mutations Spectrum in Iranian Population. Hemoglobin. 2001; 25(3):285–96.

345. **Najmabadi H**, Teimourian Sh, Khatibi T, Neishabury M, Poufrafzad F, Jalil Negad S, Azad M, Oberkanins Ch, Krugluger W. Comparison of Amplification Refractory System (ARMS) and Reverse Hybridization for the detection of the 10 most common Beta-Thalassemia mutations within the Iranian Population. Arch Irn Med. 2001; 4(4):165-170.

346. Feleki X, **Najmabadi H**, Karimi-Nejad R, Christopoulos G, Kleanthous M. Identification of a novel beta0-thalassemia mutation, codons 80/81 (-C), in an Iranian family. Hemoglobin. 2000; 24(4):319-21.

347. **Najmabadi H**, Sahebjam S, Karimi-Nejad R, Saremi A, Sahebjam F, Shafeghati Y, Karimi-Nejad MH. Short Man with 46,X, Del (Yp) Del (Yq) Karyotype and More Distal Yq Deletion. Arch Irn Med. 2000; 3(4):204-6.

348. Chai NN, Zhou H, Hernandez J, **Najmabadi H**, Bhasin S, Yen PH. Structure and organization of the RBMY genes on the human Y chromosome: transposition and amplification of an ancestral autosomal hnRNPG gene. Genomics. 1998; 49(2):283-9.

349. **Najmabadi H**, Chai N, Kapali A, Subbarao MN, Bhasin D, Woodhouse E, Yen P, Bhasin S. Genomic structure of a Y-specific ribonucleic acid binding motif-containing gene: a putative candidate for a subset of male infertility. J Clin Endocrinol Metab. 1996; 81(6):2159-64.

350. **Najmabadi H,** Huang V, Yen P, Subbarao MN, Bhasin D, Banaag L, Naseeruddin S, De Kretser DM, Baker HWG, McLachlan RI, Loveland KA, Bhasin S. Substantial prevalence of microdeletions of the Y-chromosome in infertile men with idiopathic azoospermia and oligozoospermia detected using a sequence-tagged site-based mapping strategy. Journal of Clinical Endocrinology and Metabolism, 1996. 81:1347-52.

351. Taylor EW, **Najmabadi H**, Strathearn M, Jou NT, Liebling M, Rajavashisth T, Chanani N, Phung L, Bhasin S. Human stem cell factor promoter deoxyribonucleic acid sequence and regulation by cyclic 3',5'-adenosine monophosphate in a Sertoli cell line. Endocrinol. 1996; 137(12):5407-14.

352. Butto F, Ben Shachar G, **Najmabadi H**, Smith G. Massive cardiac tumor presenting as severe cyanosis in a newborn. Pediatr Cardiol. 1994; 15(2):103-4.

353. **Najmabadi H**, Rosenberg LA, Yuan QX, Reyaz G, Bhasin S. Transcriptional regulation of inhibin beta B messenger ribonucleic acid levels in TM.4 or primary rat Sertoli cells by 8-bromo-cyclic adenosine monophosphate. Mol Endocrinol. 1993; 7(4):561-9.

354. Butto F, Margraf L, Smith G, **Najmabadi H**. Aortic atresia and tricuspid atresia occurring in complete transposition of the great vessels. Pediatr Cardiol. 1993; 14(2):133-4.

355. Bhasin S, Rosenberg LA, Strathearn M, Sod- moriah UA, Krummen LA, Burger H, Albiston A, **Najmabadi H**. Trasncriptional Regulation of Inhibin Alpha and Beta B Subunit Genes by 3′,5′ , Cycilc- Adenosine Monophosphats. In: Burger H (Ed). Frontiers in Endocrinology: Inhibin and Related Proteins. 1993; pp:209-24.

356. **Najmabadi H**, Rosenberg LA, Yuan QX, Bhatia N, Albiston A, Burger H, Bhasin S. Transcriptional and posttranscriptional regulation of inhibin alpha-subunit gene expression in rat Sertoli cells by 8-bromo-3',5'-cyclic-adenosine monophosphate. Mol Endocrionol. 1993; 7(4):469-76.