



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: 11/4/2025
Personal Information		Nationality: Iranian
E-mail: hnajm12@yahoo.com E-mail: ho.Najmabadi@uswr.ac.ir Home Page: http://genetics.uswr.ac.ir/ University URL: http://www.uswr.ac.ir		Professor of Molecular and Medical Genetics, Director of Genetics Research Center (GRC), Director of National Reference Laboratory for Prenatal Diagnosis University of Social Welfare and Rehabilitation Sciences (USWR) Kodakyar Ave., Daneshju Blvd., Evin, 1985713834, Tehran, Iran Tel & Fax: (+9821) 22180138 http://genetics.uswr.ac.ir/ E-mail: hnajm12@yahoo.com

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 Pathology & 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. Ghasemi A, Hadei SJ, Salami Z, Saffar H, **Najmabadi H**, Okhovat AA. Myo-Neuropathy and Congenital Bilateral Cataract Due to a GFER Variant in an Iranian Family. *Current Genetic Medicine Reports*. 2025;13(1):1-9.
2. Rafat M, Bouraqi Y, Sisakht JM, Azarkeivan A, **Najmabadi H**, Neishabury M. Pyruvate Kinase Deficiency: An Underdiagnosed Cause of Severe Hemolytic Anemia in Iranian Population: Insights From Whole Exome Sequencing of Four Families and Screening of a Population-Specific Database. *International Journal of Laboratory Hematology*. 2025.
3. Yoshioka W, Sonehara K, Bakhshandeh M, Ogawa M, Eguchi K, Hayashi S, et al. 689PTracing ancient gene flow from Persia to Japan through a shared haplotype in GNE myopathy. *Neuromuscular Disorders*. 2025;53:105999.
4. **Najmabadi H**, Akhtarkhavari T, Shokouhian E, Arzhangi S, Kahrizi K. FSCN1 as a Candidate Gene for Syndromic Intellectual Disability? Evidence From a Recurrent Variant in an Iranian Cohort. *Clinical Genetics*. 2025.
5. Alagha P, Akhtarkhavari T, Shokouhian E, Ghodratpour F, Arzhangi S, **Najmabadi H**, et al. Investigation of the Clinical and Genetic Spectrum of PMM2-CDG: Insights from a Family with a Novel Variant and Previous Studies. *Archives of Iranian Medicine*. 2025;28(7):387.
6. Karimzadeh P, Kachuei M, **Najmabadi H**, Keramatipour M, Rezazadeh M, Tasharrofi B, et al. BCKDK gene mutations as a rare condition responsible for comorbid neurodevelopmental delay, autism, and epilepsy: a case series of four patients. *Annals of Medicine and Surgery*. 2025;87(7):4046-52.
7. Molaei N, Alagha P, Khanbazi A, Beheshtian M, Ahangari F, Dehdahsi S, et al. Genetic spectrum among Iranian individuals with neuromuscular disorders using Next-Generation Sequencing and multiple ligation-dependent probe amplification methods: An 11-year overview. 2025.
8. Fattahi Z, Shokouhian E, Peymani F, Babanejad M, Beheshtian M, Edizadeh M, et al. Improved Diagnostic Yield in Recessive Intellectual Disability Utilizing Systematic Whole Exome Sequencing Data Reanalysis. *Clinical Genetics*. 2025;107(6):612-9.
9. Najafabadi SZ, Ghorbanoghli Z, Ghaderi Z, Afroozan F, Talea A, Ahangari F, et al. Expanding the Clinical Phenotype Associated with the NIN Gene; Report of a Patient with Short Stature, Microcephaly and Hearing Loss. *Archives of Iranian Medicine*. 2025;28(5):313.
10. Kariminejad A, Pouya F, Ahangari F, Talebi S, Afroozan F, Verheijen FW, et al. Biallelic Variant in LYSET Associated With Mucopolidosis II-Like Phenotype. *American Journal of Medical Genetics Part A*. 2025:e64063.
11. Beheshtian M, Nouri MM, Ahangari F, Makvand M, Salmani B, Kariminejad A, et al. First Iranian Family with a Novel Missense Variant in MYO9B Gene Causing Charcot–Marie–Tooth Disease. *Archives of Iranian Medicine*. 2025;28(4):236.
12. Shamsian BS, Momtazmanesh N, Molaei Tavana P, Kazemi Aghdam M, Malek F, Shirvani A, et al. Emerging Evidence of BRAFV600E in LCH: The Iranian Experience. *Iranian Journal of Blood and Cancer*. 2025;17(1):13-9.
13. Ghasemi M, Mohseni M, Fattahi Z, Edizadeh M, Beheshtian M, Keshavarzi F, et al. Haplogroup Structure and Genetic Variation Analyses of Mitochondrial Genome SNPs in the Iranian Population. *Archives of Iranian Medicine*. 2025;28(3):140.
14. Chekroun I, Shenbagam S, Almarri MA, Mokrab Y, Uddin M, Alkhnbashi OS, et al. Genomics of rare diseases in the Greater Middle East. *Nature Genetics*. 2025;57(3):505-14.
15. Mohseni M, Ashrafi FZ, Abbaspour Rodbane E, Mokabber H, Vafaei M, Nobakht R, et al. Unraveling the Genetic Landscape of Hearing Loss: A Comprehensive Study of Azeri Families in Ardabil, Iran. *Molecular Genetics & Genomic Medicine*. 2025;13(2):e70080.
16. Kazemi N, Rezvani Rezvande R, Zare Ashrafi F, Shokouhian E, Edizadeh M, Booth KT, et al. A Frameshift Variant in ANKRD24 Implicates Its Role in Human Non-Syndromic Hearing Loss. *Clinical Genetics*. 2025;107(2):214-8.
17. Shokouhian E, Kahrizi K, **Najmabadi H**, Babanejad M. Genetic etiology of Perrault syndrome in Iranian families: first report from Iran and literature review. *Journal of Applied Genetics*. 2025:1-11.
18. Khanbazi A, Ashrafi FZ, Kelishomi MA, Rezvande R, Azad M, Ahangari F, et al. P251: New insights into the genetic landscape of DMD gene mutations in Iran and the applicability of exon skipping therapies. *Genetics in Medicine Open*. 2025;3.

19. Alagha P, Molaei N, Beheshtian M, Ahangari F, Fadaee M, Ashki M, et al. P279: Application of next-generation sequencing in the diagnosis of inherited metabolic disorders in Iran: An 11-year overview. *Genetics in Medicine Open*. 2025;3.
20. Molaei N, Alagha P, Khanbazi A, Beheshtian M, Ahangari F, Fadaee M, et al. P159: Gene and mutation spectrum among Iranian individuals with neuromuscular disorders: An 11-year consolidated overview. *Genetics in Medicine Open*. 2025;3:102124.
21. Bazazzadegan N, Babanejad M, Banihashemi S, Arzhanghi S, Kahrizi K, Booth KT, et al. A Novel Candidate Gene MACF1 is Associated with Autosomal Dominant Non-syndromic Hearing Loss in an Iranian Family. *Archives of Iranian Medicine*. 2025;28(1):63.
22. Khanbazi A, Beheshtian M, Azad M, Akbari Kelishomi M, Afroozan F, Fatehi F, et al. Comprehensive copy number analysis of spinal muscular atrophy among the Iranian population. *Scientific Reports*. 2024;14(1):1-11.
23. Kopp J, Lyubenova H, Sachsenheimer T, Kuechler O, Koch L, Glaser V, et al., editors. Biallelic loss-of-function variants in SGMS1 cause a novel metabolic disorder. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
24. Fatehi F, Ghorbanoghli Z, Kooshki M, Najafabadi SZ, Noudehi K, Makvand M, et al., editors. Homozygous variant in NRDC gene in two siblings with developmental delay and seizure. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
25. Ghorbanoghli Z, Ahangari F, Afroozan F, Ramezani M, **Najmabadi H**, Kariminejad A, editors. B3GALNT2-Related Dystroglycanopathy: Expanding the Phenotype. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
26. Najafabadi SZ, Ghaderi Z, Ghorbanoghli Z, Fatehi F, Ahangari F, **Najmabadi H**, et al., editors. Expanding the clinical phenotype of patient with homozygous variant in NIN gene. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
27. Tahmasebivand M, Ghodrattpour F, Ashrafi FZ, Mohseni M, Arzhanghi S, Riazalhosseini Y, et al., editors. Contribution of four novel variants with inherited familial coronary artery disease. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
28. Rezvande RR, Kazemi N, Ashrafi FZ, Edizadeh M, Ghodrattpour F, Keshavarzi F, et al., editors. Whole-exome sequencing revealed a novel candidate gene, ARHGAP22, as potential cause of non-syndromic hearing loss in an Iranian family. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
29. Molaei N, Ahangari F, Beheshtian M, Alagha P, Dehdahsi S, Fadaie M, et al., editors. The utility of whole exome sequencing in accurate genetic diagnosis of neuromuscular disorders in Iran. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
30. Ghasemi M, Mohseni M, Edizadeh M, Riazalhosseini Y, Kahrizi K, **Najmabadi H**, editors. Haplogroup structure and genetic variation analyses of complete mtDNA genomes in Iranian ethnolinguistic population. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
31. Alagha P, Ahangari F, Beheshtian M, Molaei N, Dehdahsi S, Fadaie M, et al., editors. A high diagnostic rate of inherited metabolic disorders by whole exome sequencing. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
32. Kazemi N, Rezvande RR, Ashrafi FZ, Ghodrattpour F, Keshavarzi F, Jalalvand K, et al., editors. DBX2: A novel candidate gene for non-syndromic hearing loss identified in an Iranian family. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.
33. Ahangari F, Dehdahsi S, Fadaie M, Ashki M, Beheshtian M, Azad M, et al., editors. Implementation of Expanded Carrier Screening for Genetic Diseases: Report from Iran. *EUROPEAN JOURNAL OF HUMAN GENETICS*; 2024: SPRINGER NATURE CAMPUS, 4 CRINAN ST, LONDON, N1 9XW, ENGLAND.

34. Reshadmanesh A, Dehdahsi S, Ahangari F, Kahrizi K, Kariminejad A, Mahdavi SS, et al. First Case of Macrocephaly, Dysmorphic Facies, and Psychomotor Retardation Harboring Co-inherited Variants in HERC1 and PMP22 Genes from Iran: Two Novel Variants. *Archives of Iranian Medicine*. 2024;27(12):700.
35. Ghasemi, A., Hadei, S.J., KamaliZonouzi, S., Shahrokhi, A., **Najmabadi, H.** and Nafissi, S., 2025. Clinical and genetic diversity in Iranian individuals with RAPSN-related congenital myasthenic syndrome. *Neurogenetics*, 26(1), pp.1-11.
36. Ashrafi, F.Z., Dorgaleleh, S., Rezvandeh, R.R., Kazemi, N., Azizi, N., Edizadeh, M., Azizi, M.H., Kahrizi, K., **Najmabadi, H.**, Najafipour, R. and Mohseni, M., 2024. A Village in the Southeastern Region of Iran Harboring the c. 716T> A (p. Val239Asp) Mutation in SLC26A4. *Archives of Iranian Medicine*, 27(9), p.522.
37. Khoeini, T., Kariminejad, A., Nilipour, Y., Ariaei, A., **Najmabadi, H.**, Arabshahi, M., Faraji Zonooz, M. and Haghi Ashtiani, B., 2024. Core myopathy in two siblings with a biallelic variant in the CACNA1S gene—A case series study. *Clinical Case Reports*, 12(8), p.e9251.
38. Zarifian Yeganeh, R., Akbari Kelishomi, M., Ahmadpour Jenaghari, A., Salmani, B., Vahidi, Z., Makvand, M., Azad, M., Kooshki, M., Bouraqi, Y., Azarkeivan, A. and **Najmabadi, H.**, 2024. HFE and Non-HFE Hereditary Hemochromatosis Based on Screening of 854 Individuals: 12 Years of an Iranian Experience. *Genetic Testing and Molecular Biomarkers*, 28(7), pp.289-296.
39. Mehvari, S., Karimian Fathi, N., Saki, S., Asadnezhad, M., Arzhanghi, S., Ghodrattpour, F., Mohseni, M., Zare Ashrafi, F., Sadeghian, S., Boroumand, M. and Shokohizadeh, F., 2024. Contribution of genetic variants in the development of familial premature coronary artery disease in a cohort of cardiac patients. *Clinical Genetics*, 105(6), pp.611-619.
40. Alavi, S., Mohammadimoghaddam, S., **Najmabadi, H.** and Maghsoudlou, S., 2024. The First Iranian Case of Unstable Hemoglobin Santa Ana. *Hemoglobin*, 48(2), pp.125-128.
41. Abolhassani, A., Fattahi, Z., Beheshtian, M., Fadaee, M., Vazehan, R., Ahangari, F., Dehdahsi, S., Faraji Zonooz, M., Parsimehr, E., Kalhor, Z. and Peymani, F., 2024. Clinical application of next generation sequencing for Mendelian disease diagnosis in the Iranian population. *NPJ Genomic Medicine*, 9(1), p.12.
42. Alinaghi S, Mohseni M, Fattahi Z, Beheshtian M, Ghodrattpour F, Ashrafi FZ, Arzhanghi S, Jalalvand K, Najafipour R, Khorshid HR, Kahrizi K. Genetic Analysis of 27 Y-STR Haplotypes in 11 Iranian Ethnic Groups. *Archives of Iranian Medicine*. 2024 Feb;27(2):79.
43. Kazemi, N., Rezvani Rezvandeh, R., Zare Ashrafi, F., Shokouhian, E., Edizadeh, M., Booth, K.T., Kahrizi, K., Najmabadi, H. and Mohseni, M., 2024. A Frameshift Variant in ANKRD24 Implicates Its Role in Human Non-Syndromic Hearing Loss. *Clinical Genetics*.
44. Mehvari S, Fathi NK, Asadnezhad M, Arzhanghi S, Sadeghian S, Boroumand M, et al. P292: Contribution of rare variants in the development of familial premature coronary artery disease in a cohort of cardiac patients. 2024;2.
45. Abolhassani A, Fattahi Z, Beheshtian M, Fadaee M, Vazehan R, Ahangari F, et al. P604: Diagnostic utility of NGS testing in a highly consanguineous population: Findings from 1400+ Iranian patients with Mendelian disorders. 2024;2.
46. Khanbazi A, Beheshtian M, Azad M, Akbari M, Afrozani F, Fatehi F, et al. P483: A comprehensive study of spinal muscular atrophy testing referrals' data among the Iranian population since 2006. 2024;2.
47. 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*, 614(7948), pp.564-571.
48. Rashvand, Z., **Najmabadi, H.**, Kahrizi, K., Mozhdehipanah, H., Moradi, M., Estaki, Z., Taherkhani, K., Nikzat, N., Najafipour, R. and Omrani, M.D., 2024. 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*, 18(1), p.25.
49. Mahmoudi-Aznaveh, A., Tavoosidana, G., **Najmabadi, H.**, Azizi, Z. and Ardestani, A., 2023. The liver-derived exosomes stimulate insulin gene expression in pancreatic beta cells under condition of insulin resistance. *Frontiers in Endocrinology*, 14, p.1303930.

50. Bazazzadegan, N., Abedini, S.S., Azarkeivan, A., Banihashemi, S., Nikzat, N., **Najmabadi, H.** and Neishabury, M., 2023. The Spectrum of HBB Mutations among 2315 Beta Thalassemia Patients of a Reference Clinic in Tehran-Iran. *Hemoglobin*, 47(4), pp.147-151.
51. 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*, 104(1), pp.121-126.
52. Moghadam, M.G., Elahi, Z., Soveyzi, M., Arzhangi, S., Nafissi, S., **Najmabadi, H.**, Kahrizi, K. and 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*, 26(5), p.279.
53. Jamshidi, F., Shokouhian, E., Mohseni, M., Kahrizi, K., **Najmabadi, H.** and 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), p.e2168.
54. Ashrafi, F.Z., Akhtarkhvari, T., Fattahi, Z., Asadnezhad, M., Beheshtian, M., Arzhangi, S., **Najmabadi, H.** and 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), p.186.
55. 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*, 11(4), p.e2131.
56. Mohseni, M., Mohammadi, Y., Ashrafi, F.Z., Ghodrattpour, F., Jalalvand, K., Arzhangi, S., Babanejad, M., Azizi, M.H., Kahrizi, K. and **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), p.176.
57. 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*, 191(3), pp.794-804.
58. 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*, 614(7948), pp.564-571.
59. Ashrafi, F.Z., Mohseni, M., Beheshtian, M., Fattahi, Z., Ghodrattpour, F., Keshavarzi, F., Behravan, H., Kalhor, M., Jalalvand, K., Azad, M. and Koshki, M., 2023. Implementation of an in-house platform for rapid screening of SARS-CoV-2 genome variations. *Archives of Iranian medicine*, 26(2), p.69.
60. Mahmoudi-Aznaveh, A., Tavoosidana, G., **Najmabadi, H.**, Azizi, Z. and Ardestani, A., 2023. The liver-derived exosomes stimulate insulin gene expression in pancreatic beta cells under condition of insulin resistance. *Frontiers in Endocrinology*, 14, p.1303930.
61. Ehtesham, N., Mosallaei, M., Beheshtian, M., Khoshbakht, S., Fadaee, M., Vazehan, R., Zonooz, M.F., Karimzadeh, P., Kahrizi, K. and **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), p.788.
62. Saghi, M., InanlooRahatloo, K., Alavi, A., Kahrizi, K. and **Najmabadi, H.**, 2022. Intellectual disability associated with craniofacial dysmorphism due to POLR3B mutation and defect in spliceosomal machinery. *BMC Medical Genomics*, 15(1), p.89.
63. Sisakht, J.M., Mehri, M., **Najmabadi, H.**, Azarkeivan, A. and 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), p.691.
64. Hosseinpour, M., Ardalani, F., Mohseni, M., Beheshtian, M., Arzhangi, S., Ossareh, S., **Najmabadi, H.**, Nobakht, A., Kahrizi, K. and 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), p.600.

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66. Karimzadeh, P., **Najmabadi, H.**, Lochmuller, H., Babaee, M., Dehdahsi, S., Miryounesi, M., Amirsalari, S., Rayegani, S.M. and 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*, 32(10), pp.806-810.
67. Goodarzi, A., Rad, F., Esmaeili, O.S., Forouzeshpour, F., **Najmabadi, H.** and Azarkeivan, A., 2022. 3091–CLINICAL FEATURES AND MOLECULAR BASIS OF THALASSEMIC PATIENTS WITH HOMOZYGOTE FORM OF IVSII-IVSII-I MUTATION AND SYNCHRONIZATION WITH A-GENE MUTATION. *Experimental Hematology*, 111, p.S90.
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69. Fattahi, Z., Mohseni, M., Jalalvand, K., Aghakhani Moghadam, F., Ghaziasadi, A., Keshavarzi, F., Yavarian, J., Jafarpour, A., Mortazavi, S.E., Ghodrattpour, F. and Behravan, H., 2022. SARS-CoV-2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. *Transboundary and emerging diseases*, 69(3), pp.1375-1386.
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