University of Social Welfare and Rehabilitation Sciences
Ministry of Health and Medical Education

ITA		
Name: Kimia Surname: Kahrizi	Specialty /Ph.D. Medical Doctor, Pediatrician, Clinical geneticist Professor of Medical Genetics	
Title/Degree: Prof.	Department of: Genetics	

Research Interests:

- Genetic investigation of hereditary disorders for instances intellectual disability, hereditary microcephaly, hereditary hearing loss, dysmorphology and recently investigation on familial premature coronary artery diseases.

Scopus Profile: https://www.scopus.com/authid/detail.uri?authorId=12242319600 Google Scholar Profile: https://scholar.google.com/scholar?hl=en&as_sdt=0%2C5&q=Kahrizi+Kimia&btnG=	Updated:

Personal Information	**Nationality: Iranian**
Office address: Department of Genetics University of Social Welfare & Rehabilitation Sciences Kodakyar St., Daneshjo Blvd., Evin, Tehran-Iran Tel: (21) 22180138 Fax: (21) 22180138	E-mail: kkahrizi @uswr.ac.ir Home Page: https://isid.research.ac.ir/Kimia_Kahrizi University URL: http://www.uswr.ac.ir kahrizi@yahoo.com

Education

Date	Degree	Duration	Institution	Country/City	Major
1986-1994	**Medical Doctor**	6 years	Iran University of Medical Sciences	Iran/Tehran	
1994 -1997	**Pediatrics**	3 years	Iran University of Medical Sciences	Iran/Tehran	
1998- 1998	**Deputy for Preventive Affairs**	1 year	Welfare Organization	Iran/Tehran	

Faculty member

Year	Position	Duration	Institution/Course	Location
1999- 2006	**Assistant Professor**	7 years	Dep: Genetics Research Center; University of Social Welfare & Rehabilitation Sciences	
2006- 2011	**Associate Professor**	5 yeras	Dep: Genetics Research Center; University of Social Welfare & Rehabilitation Sciences	
2011-present	**Professor**	10 years	Dep: Genetics Research Center; University of Social Welfare & Rehabilitation Sciences	

Field of Specialization

A talented medical researcher with a significant involvement in research projects in the field of Neuromuscular Disorders such as Myotonic Dystrophy, Limb Girdle Muscular Dystrophy (LGMD), Duchene Muscular Dystrophy (DMD) and Congenital Muscular Dystrophy (CMD), Hereditary Hearing Loss (HHL) including Usher, Pendred, and Distal renal tubular acidosis (dRTA), and Intellectual Disability (ID), syndromic microcephaly and brain malformations and also expertise in diagnosis of dysmorphic patients as well as novel syndromes. Recently involved in genetic investigation of familial premature coronary disease.Fifteen years experience in deep phenotyping to phenotype-genotype matching of exome sequencing data in research and diagnostic sectors.

Language Ability

- English

- Persian

-French

Research Experience

Year	Position	Institution/Course	Location

Scientific Membership

Year	Association, Society	Location
Annually	European Human Genetic Society	Euro
Annually	Iranin Neurogenetic Society	Iran
Annually	Iranian Human Genetic Society	Iran

Publications

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.

Mehvari, S., Karimian Fathi, N., Saki, S., Asadnezhad, M., Arzhangi, S., Ghodratpour, 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.

Rashvand, Z., Najmabadi, H., **Kahrizi, K**., (...), Najafipour, R., Omrani, M.D. 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. 18(1), pp. 25-41.

Jamshidi, F., Shokouhian, E., Mohseni, M., **Kahriz, K.,** Najmabadi, H., Babanejad, M. 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 and Genomic Medicine, 2023.11(5),e2168.

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

Mensah, M.A., Niskanen, H., Magalhaes, A.P(...), **Kahrizi., K.,** (...), Horn, D., Hnisz, D. Aberrant phase separation and nucleolar dysfunction in rare genetic diseases. Nature,2023. 614(7948), pp. 564-571

Ashrafi, F.Z., Mohseni, M., Beheshtian, M., (...), **Kahrizi, K**., Jazayeri, S.M., Najmabadi, H.Implementation of an In-House Platform for Rapid Screening of SARS-CoV-2 Genome Variations. Archives of Iranian Medicine,2023. 26(2), pp. 69-75

Moghadam, M.G., Elahi, Z., Soveyzi, M., (...), **Kahrizi, K**., Fattahi, Z. Expanding the Molecular Spectrum of HK1-Related Charcot-Marie-Tooth Disease, Type 4G; the First Report in Iran. Archives of Iranian Medicine, 2023. 26(5), pp. 279-284

Ashrafi, F.Z., Akhtarkhavari, T., Fattahi, Z., (...), Najmabadi, H., **Kahrizi, K.** Emerging Epidemiological Data on Rare Intellectual Disability Syndromes from Analyzing the Data of a Large Iranian Cohort. Archives of Iranian Medicine, 2023. 26(4), pp. 186-197

Dawood, M., Akay, G., Mitani, T., Marafi, D., Fatih, J. M., Gezdirici, A., Najmabadi, H**., Kahrizi, K**., (...), Gibbs, R.A., Lupski, J.R. A biallelic frameshift indel in PPP1R35 as a cause of primary microcephaly. American Journal of Medical Genetics, 2023. Part A, 191(3), pp. 794-804.

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

Ehtesham, N., Mosallaei, M., Beheshtian, M., (...), **Kahrizi, K**., Najmabadi, H. Characterizing Genotypes and Phenotypes Associated with Dysfunction of Channel-Encoding Genes in a Cohort of Patients with Intellectual Disability. Archives of Iranian Medicine, 2022. 25(12), pp. 788-797

Divya Nair,Dong Li, Hannah Erdogan, Andrew Yoon, Margaret H. Harr, Gaber Bergant, Borut Peterlin, Marusa Skrjanec Pusenjak, Parul Jayakar, Rolph Pfundt, Sandra Jansen,4 Kirsty McWalter, Alpa Sidhu, Sheila Saliganan, Emanuele Agolini,8 Arthur Jacob, Jennifer Pasquier, Rafii Arash, **Kimia Kahrizi**, Hossein Najmabadi, Hans-Hilger Ropers, and Elizabeth J. Bhoj1,* Discovery of a neuromuscular syndrome caused by biallelic variants in *ASCC3*. Human Genetics and

Genomics Advances, 2022. 2, 100024, April 8.

Hosseinpour, M., Ardalani, F., Mohseni, M., (...), **Kahrizi, K.**, Broumand, B. 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, 2022. 25(9), pp. 600-608

Fattahi, Z., Mohseni, M., Beheshtian, M., (...), **K. Kahrizi.,** Jazayeri, S.M., Najmabadi, H. Disease Waves of SARS-CoV-2 in Iran Closely Mirror Global Pandemic Trends. Archives of Iranian Medicine, 2022. 25(8), pp. 508-522.

Sumathipala, D. Strømme, P., Fattahi, Z., Lüders, T., Sheng, Y., **Kahrizi, K**., Holm Einarsen, I., Sloan, J.L., Najmabadi, H., (...), Frengen, E., Misceo, D. ZBTB11 dysfunction: Spectrum of brain abnormalities, biochemical signature and cellular consequences. Brain, 2022. 145(7), pp. 2602-2616.

Fattahi, Z., Mohseni, M., Jalalvand, K., (...), **Kahrizi, K**., Jazayeri, S.M., Najmabadi, H. SARS-CoV-2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. Transbound Emerg Dis. 2022 Apr 9;10.1111/tbed.14104

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

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.

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.

Monika Oláhová , Bradley Peter , Zsolt Szilagyi , Hector Diaz-Maldonado, Meenakshi Singh, Ewen W Sommerville , Emma L Blakely , Jack J Collier , Emily Hoberg , Viktor Stránecký , Hana Hartmannová , Anthony J Bleyer , Kim L McBride , Sasigarn A Bowden , Zuzana Korandová , Alena Pecinová , Hans-Hilger Ropers , **Kimia Kahrizi** , Hossein Najmabadi , (…) , Claes M Gustafsson, Robert W Taylor . POLRMT mutations impair mitochondrial transcription causing neurological disease. Nat Commun. 2021. Feb 18;12(1):1135.

Averdunk, L., Sticht, H., Surowy, 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

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

Mohseni, M., Babanejad, M., Booth, K.T., (...), **Kahrizi, K**., Najmabadi, H. Exome sequencing utility in

defining the genetic landscape of hearing loss and novel-gene discovery in Iran. Clinical Genetics,2021. 100(1), pp. 59-78.

Sheikh, T.I., Vasli, N., Pastore, S., **Kahrizi, K**., (...), Villunger, A., Vincent, J. B.Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. Translational Psychiatry. 2021, 11(1),1.

Khoshbakht, S., Beheshtian, M., Fattahi, Z., (...), **Kahrizi, K**., Najmabadi, H. CEP104 and CEP290; Genes with ciliary functions cause intellectual disability in multiple families. Arch Iran Med. 2021;24(5): 364-373.

Beheshtian, M., Akhtarkhavari, T., Mehvari, S., (...), Najmabadi, H., **Kahrizi, K**. Comprehensive genotype-phenotype correlation in AP-4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clinical Genetics, 2021. 99(1), pp. 187-192.

Rashvand, Z., **Kahrizi, K**., Najmabadi, H., Najafipour, R., Omrani, M.D. Clinical and genetic characteristics of splicing variant in cyp27a1 in an iranian family with cerebrotendinous xanthomatosis.Iranian Biomedical Journal, 2021 Mar. 25(2), pp. 132-139.

Kazemi, G., Peymani, F., Mohseni, M., (...), **Kahrizi, K**., Najmabadi, H. Novel mutation in LARP7 in two Iranian consanguineous families with syndromic intellectual disability and facial dysmorphism. Archives of Iranian Medicine2020, 23(12), pp. 842-847.

Jorge Diaz, Xavier Gérard, Michel-Boris Emerit, Julie Areias, David Geny, Julie Dégardin, Manuel Simonutti, Marie-Justine Guerquin, Thibault Collin, Cécile Viollet, Jean-Marie Billard, Christine Métin, Laurence Hubert, Farzaneh Larti, **Kimia Kahrizi**, Rebekah Jobling, (…), Susan Blaser, Maha Gashlan, Claude Besmond, Michèle Darmon, Justine Masson. YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. *Brain*, Volume 143, Issue 10, October 2020, Pages 2911–2928.

Sepideh Mehvari, Farzaneh Larti, Hao Hu, Zohreh Fattahi, Maryam Beheshtian, Seyedeh Sedigheh Abedini, Sanaz Arzhangi, Hans-Hilger Ropers, Vera M. Kalscheuer, Daniel Auld**, Kimia Kahrizi**, Yasser Riazalhosseini, Hossein Najmabadi. Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics and Genomic Medicine, Oct. 2020. 8(10),e1418

Taghizadeh, S., Vazehan, R., Beheshtian, M., (...), Najmabadi, H., **Kahrizi, K**.Molecular diagnosis of hereditary neuropathies by whole exome sequencing and expanding the phenotype spectrum. 2020. Archives of Iranian Medicine, 23(7):426-433

Mohseni, M., Akbari, M., Booth, K.T., (...), **Kahrizi, K**., Najmabadi, H. When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). Journal of Human Genetics,2020. no.7 (2020): 609.0-617.0.

Mehrjoo, Z., **Kahrizi, K**., Mohseni, M., (...), Mohebbi, S., Daneshi, A. Limbic system associated membrane protein mutation in an iranian family diagnosed with ménière's disease. Archives of Iranian Medicine

2020, 23(5), pp. 319-325.

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. European Journal of Medical Genetics, April 2020, 63(4),103849.

Mitani, T., Punetha, J., Akalin, I., (...), **Kahrizi, K**., Gawlinski, P. Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics,Nov. 2019, 105(5), pp. 1005-1015.

Zohreh Fattahi, Maryam Beheshtian , Marzieh Mohseni , Hossein Poustchi, Erin Sellars, Sayyed Hossein Nezhadi, Amir Amini , Sanaz Arzhangi, Khadijeh Jalalvand, Peyman Jamali, Zahra Mohammadi, Behzad Davarnia , Pooneh Nikuei, Morteza Oladnabi, Akbar Mohammadzadeh, Elham Zohrehvand, Azim Nejatizadeh, Mohammad Shekari, Maryam Bagherzadeh, Ehsan Shamsi-Gooshki , Stefan Börno, Bernd Timmermann, Aliakbar Haghdoost, Reza Najafipour, Hamid Reza Khorram Khorshid, **Kimia Kahrizi**, Reza Malekzadeh, Mohammad R Akbari , Hossein Najmabadi. Iranome: A catalog of genomic variations in the Iranian population. Hum Mutat, 2019 Nov;40(11):1968-1984

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

Ataei, R., Khoshbakht, S., Beheshtian, M., (...), **Kahrizi, K**., Najmabadi, H. Contribution of Iran in Elucidating the Genetic Causes of Autosomal Recessive Intellectual Disability. Arch Iran Med. 2019 Aug 1;22(8):461-471

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 Jul;24(7):1027-1039.

Kahrizi, K., Huber, M., Galetzka, D., (...), Najmabadi, H., Winter, J. Homozygous variants in the gene SCAPER cause syndromic intellectual disability.American Journal of Medical Genetics, 2019. Part A, 179(7), pp. 1214-1225.

Beheshtian, M., Fattahi, Z., Fadaee, M., (...), 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 Jun;95(6):718-725.

Hosseini, M., Larti, F., Fattahi, Z., Najmabadi, H., **Kahrizi, K**. A splice-altering variant in LARP7 gene leads to exon exclusion. Gene Reports. Volume 15, June 2019, 100375

Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability PatientsInanlooRahatloo, K., Peymani, F., **Kahrizi, K**., Najmabadi, H. Neuroscience. 2019 Apr15;404:423-444

Mehregan, H., Mohseni, M., Akbari, M., (...), Kahrizi, K., Najmabadi, H. Novel mutations in KCNQ4, LHFPL5 and COCH genes in iranian families with hearing impairment. Arch Iran Med. 2019 Apr 1;22(4):189-197.

Mehregan, H., Mohseni, M., Jalalvand, K., (...), **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 Feb;117:115-126.

Kahrizi, K., Hu, H., Hosseini, M., (...), Najmabadi, H., Ropers, H.-H. Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics , 2019. 95(1), pp. 151-159.

Hosseini, M., Fattahi, Z., Abedini, S.S., (...), Najmabadi, H., **Kahrizi, K**. GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. 2019 Jan;179(1):13-19

Zohreh Mehrjoo, Z. Fattahi, M. Beheshtian, M. Mohseni, H. Poustchi, Fariba Ardalani, K. Jalalvand, S. Arzhangi, Zahra Mohammadi, Shahrouz Khoshbakht, F. Najafi, P. Nikuei, M. Haddadi, Elham Zohrehvand, M. Oladnabi, Akbar Mohammadzadeh, M. Jafari, T. Akhtarkhavari, E. S. Gooshki, A. Haghdoost, R. Najafipour, Lisa-Marie Niestroj, B. Helwing, Yasmina Gossmann, M. Toliat, R. Malekzadeh, P. Nürnberg, **K. Kahrizi**, H. Najmabadi, M. Nothnagel. Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genet. 2019 Sep 24;15(9):e1008385

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 Dec;177(8):691-699

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 Nov;94(5):450-456.

Abedini SS, Kahrizi K, de Pouplana LR, Najmabadi H. tRNA Methyltransferase Defects and Intellectual Disability. Arch Iran Med. 2018 Oct 1;21(10):478-485.

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 Sep 15;27(18):3177-3188.

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

Aug;55(8):555-560.

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 K, Naz S, Friedman TB. CDC14A Phosphatase is Essential for Hearing and Male Fertility in Mouse and Human. Human Molecular Genetics,2018. 27(5), pp. 780-798

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 FA, 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. Molecular Psychiatry, 2018. volume 23, pages 222–230

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. 2017 Nov 7.

Hosseini M, Najmabadi H, **Kahrizi K**. Calpains: Diverse Functions but Enigmatic. Arch Iran Med. 2018 Apr 1;21(4):170-179.Review PMID:29693408

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. 2017 Dec 19.

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 Jan;104:195-199.

Kazeminasab S, Najmabadi H, Kahrizi K. Intellectual Disability and Ataxia: Genetic Collisions. Archives of Iranian Medicine, 21(1), pp. 29-40

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 Dec;9(12):1711-1731.

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 Sep 7;101(3):428-440.

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 Sep;31(3):161-169.

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 May 26;3(3):e148.

Musante L, Püttmann L, **Kahrizi K**, Garshasbi M, Hu H, Stehr H, Lipkowitz B, Otto S, Jensen LR, Tzschach A, Jamali P, Wienker T, Najmabadi H, Ropers HH, Kuss AW. Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. Hum Mutat. 2017 Jun;38(6):621-636.

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. Clinical Genetics, 91(3):386-402

Ostaresh, F., Hadji-Alikhani, R., Babanejad, M., (...), Najmabadi, H., **Kahrizi, K**. Linkage analysis for four genes (GPSM2, MSRB3, SLC26A5,GRXCR1) in 100 Iranian families with autosomal recessive hearing loss. Genetics in the Third Millennium,2016, 13(4), pp. 4094-4099.

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 Oct 1;19(10):720-728.

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 Sep;95(3):667-74.

Mehregan H, Najmabadi H, **Kahrizi K**. Genetic Studies in Intellectual Disability and Behavioral Impairment. Arch Iran Med. 2016 May;19(5):363-75.

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 May 11;12(5):e1006022.

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 Apr-May;26(4-5):277-82.

Staufner C, Lindner M, Dionisi-Vici C, Freisinger P, Dobbelaere D, Douillard C, Makhseed N, Straub BK, **Kahrizi K**, Ballhausen D, la Marca G, Kölker S, Haas D, Hoffmann GF, Grünert SC, Blom HJ. Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. J Inherit Metab Dis. 2016 Mar;39(2):273-83.

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 Nov;18(11):776-85.

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 Dec;52(12):823-9.

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 Oct;18(10):688-97

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 Oct;18(10):670-82.

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 Oct;18(10):643-69.

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 Sep;94(3):483-7.

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 Dec;167A(12):2957-65.

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 Aug 26;10(8):e0129631

Iqbal Z, Püttmann L, Musante L, Razzaq A, Zahoor MY, Hu H, Wienker TF, Garshasbi M, Fattahi Z, Gilissen C, Vissers LE, de Brouwer AP, Veltman JA, Pfundt R, Najmabadi H, Ropers HH, Riazuddin S, **Kahrizi K**, van Bokhoven H. Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. Eur J Hum Genet. 2016 Mar;24(3):392-9.

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 Mar;18(3):179-84.

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, D'heedene 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 Mar 11.

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 Mar 5;96(3):386-96.

Larti F, **Kahrizi K**, Musante L, Hu H, Papari E, Fattahi Z, Bazazzadegan N, Liu Z, Banan M, Garshasbi M, Wienker TF, Hilger Ropers H, Galjart N, Najmabadi H. A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. Eur J Hum Genet. 2015 Mar;23(3):416.

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 Dec;93(3):837-41. No abstract available.

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 Jan;18(1):60-4

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 Feb;79(2):136-8

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 Dec;128(12):1056-9

Najmabadi H, **Kahrizi K.** Genetics of non-syndromic hearing loss in the Middle East. Int J Pediatr Otorhinolaryngol. 2014 Dec;78(12):2026-2036. doi: 10.1016/j.ijporl.2014.08.036. Epub 2014 Sep 4. Review.

Shearer AE, Eppsteiner RW, Booth KT, Ephraim SS, Gurrola J 2nd, 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 Oct 2;95(4):445-53.

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 Nov;78(11):1828-1832

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 Sep 13.

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 Aug 14

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 May;52(5):352-9.

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 nine novel mutations in five BBS genes. J Hum Genet. 2014 May 22

Khoshbakht R, Soltanzadeh A, Zamani B, Abdi S, Gharagozli K, **Kahrizi K**, Khoshbakht R, Nafissi S. J Clin Neurosci. 2013 Nov 18. Correlation between distribution of muscle weakness, electrophysiological findings and CTG expansion in myotonic dystrophy.

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 Spring;7(2):23-30.

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 Oct;15(10):e8221

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

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 Nov;62(11):1244-7.

Püttmann L, Stehr H, Garshasbi M, Hu H, **Kahrizi K**, Lipkowitz B, Jamali P, Tzschach A, Najmabadi H, Ropers HH, Musante L, Kuss AW. A novel ALDH5A1 mutation is associated with succinic

semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. Am J Med Genet A. 2013 Aug;161A(8):1915-22.

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. doi: 10.1371/journal.pone.0053826. Epub 2013 Jan 9.

Papari E, Bastami M, Farhadi A, Abedini S, Hosseini M, Bahman I, Mohseni M, Garshasbi M, Moheb LA, Behjati F, **Kahrizi K**, Ropers HH, Najmabadi H. Investigation of primary microcephaly in Bushehr province of Iran: novel STIL and ASPM mutations. Clin Genet. 2013 May;83(5):488-90.

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 Oct;158A(10):2485-92. doi: 10.1002/ajmg.a.35572. Epub 2012 Aug 17.

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 Feb;14(2):79-85. Epub 2012 Feb 1.

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 Aug;158A(8):1857-64. doi: 10.1002/ajmg.a.34411. Epub 2012 Jun 26.

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 Aug;76(8):1164-74. Epub 2012 Jun 12.

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 Jun;15(6):361-5.

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 Apr 26.

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 Apr;7(2):321-5. Epub 2011 May 17.

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. 2011 Dec 13.

Najmabadi H, Hu H, Garshasbi M, Zemojtel T, Abedini SS, Chen W, Hosseini M, Behjati F, Haas S, Jamali P, Zecha A, Mohseni M, Püttmann L, Vahid LN, Jensen C, Moheb LA, Bienek M, Larti F, Mueller I, Weissmann R, Darvish H, Wrogemann K, Hadavi V, Lipkowitz B, Esmaeeli-Nieh S, Wieczorek D, Kariminejad R, Firouzabadi SG, Cohen M, Fattahi Z, Rost I, Mojahedi F, Hertzberg C, Dehghan A, Rajab A, Banavandi MJ, Hoffer J, Falah M, Musante L, Kalscheuer V, Ullmann R, Kuss AW, Tzschach A, **Kahrizi K**, Ropers HH. Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature. 2011 Sep 21;478(7367):57-63.

Norouzi V, Azizi H, Fattahi Z, Esteghamat F, Bazazzadegan N, Nishimura C, Nikzat N, Jalalvand K, **Kahrizi K**, Smith RJ, Najmabadi H. Did the GJB2 35delG mutation originate in Iran? Am J Med Genet A. 2011 Sep 9.

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 Sep 9;89(3):407-14.

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 Jul 15;89(1):176-82.

Pak C, Garshasbi M, **Kahrizi K**, Gross C, Apponi LH, Noto JJ, Kelly SM, Leung SW, Tzschach A, Behjati F, Abedini SS, Mohseni M, Jensen LR, Hu H, Huang B, Stahley SN, Liu G, Williams KR, Burdick S, Feng Y, Sanyal S, Bassell GJ, Ropers HH, Najmabadi H, Corbett AH, Moberg KH, Kuss AW. Mutation of the conserved polyadenosine RNA binding protein, ZC3H14/dNab2, impairs neural function in Drosophila and humans. Proc Natl Acad Sci U S A. 2011 Jul 26;108(30):12390-5. Epub 2011 Jul 6.

Masoud Garshasbi, **Kimia Kahrizi***, Masoumeh Hosseini, Leila Nouri Vahid, Masoumeh Falah, Sahel Hemmati, Hao Hu,1 Andreas Tzschach, Hans Hilger Ropers, ** Hossein Najmabadi, and Andreas Walter Kuss. A Novel Nonsense Mutation in TUSC3 is Responsible for Non-Syndromic Autosomal Recessive Mental Retardation in a Consanguineous Iranian Family. American Journal of Medical Genetics, 2011.

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 Apr 11.

Borck G, Rehman AU, 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 Feb 11;88(2):127-37. Epub 2011 Jan 20.

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 Dec;119(12):830-5.

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.

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. 2010 Nov 10.

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

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 Dec;169(12):1535-9.

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 Nov;18(11):1178-84.

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 Mar;152A(3):646-52.

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. Mir A, Kaufman L, Noor A, Motazacker MM, Jamil T, Azam M,. Am J Hum Genet. 2009 Dec;85(6):909-15.

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

Pouya AR, Abedini SS, Mansoorian N, Behjati F, Nikzat N, Mohseni M, Nieh SE, Abbasi, Moheb 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 Jul-Aug;52(4):170-3. Epub 2009 Apr 8.

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 Apr;84(4):505-10. Epub 2009 Apr 2.

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

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 Feb;38(2):139-41.

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 Mar;149A(3):555-8. No abstract available.

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 Dec 15;146A (24): 3223-6. No abstract available.

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

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 Jun;168(6):651-3. Epub 2008 Sep 24.

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

Naghavi A, Nishimura C, **Kahrizi K**, Riazalhosseini Y, Bazazzadegan N, Mohseni M, Smith RJ, Najmabadi H. GJB2 mutations in Baluchi population. J Genet. 2008 Aug;87(2):195-7. No abstract available.

Behjati F, Shafaghati Y, Firouzabadi SG, **Kahrizi K**, Bagherizadeh I, Najmabadi H, Bint S, Ogilvie C. Eur J Med Genet. M-banding characterization of a 16p11.2p13.1 tandem duplication in a child with autism, neurodevelopmental delay and dysmorphism. 2008 Nov-Dec;51(6):608-14. Epub 2008 Jul 12.

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 May;35(2):216-9.

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 May;82(5):1158-64.

Yousef Shafeghati, M.D.; **Kimia Kahrizi**, M.D.; Hossein Najmabadi, PhD; Andreas W Kuss, PhD; Hans-Hilger Ropers, M.D.; Andreas Tzschach, M.D. Brachyphalangy, polydactyly and tibial aplasia/hypoplasia syndrome: case report and review of the literature. Eur J Pediatr (2010) 169: 1535.

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 Apr;77 (4):395-8.

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 Mar;152A(3):646-52.

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 Feb 4.

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 Dec;85(6):909-15.

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 Sep;85(3):328-37.

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. 2009 Jun 15.

Pouya AR, Abedini SS, Mansoorian N, Behjati F, Nikzat N, Mohseni M, Nieh SE, Abbasi Moheb 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 Jul-Aug;52(4):170-3.

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 Apr;46(4):272-6.

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 Apr 1. [Epub ahead of print]

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 a common cause of DFNB3 hearing loss. Laryngoscope. 2009 Mar 9.

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 Feb;38(2):139-41.

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 Feb 1;149A(3):555-8.

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 Apr;46(4):272-6.

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 Apr;84(4):505-10.

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 a common cause of DFNB3 hearing loss. Laryngoscope. 2009 Apr;119(4):727-33.

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 Feb;38(2):139-41.

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 Feb 1;149A(3):555-8. No abstract available.

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 Nov 14;146A(24):3223-3226.

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. 2008 Nov 12.

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. 2008 Sep 10.

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

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 Jul 12

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 Jun 28.

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 May;35(2):216-9.

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.

Mandana Hasanzad; Zahra Golkar; Valeh Hadavi; Roxana Kariminejad; Navid Almadani; Fariba Afroozan; Iman Salahshurifar; Yousef Shafeghati; **Kimia Kahrizi**; Hossein Najmabadi
Survival Motor Neuron Gene Deletions among Iranian Spinal Muscular Atrophy Patients
Submitted Academy of Medicine, Singapore

A, Naghavi; C. Nishimura; **K. Kahrizi**; Y. Riazalhosseini; N. Bazazzadegan; M. Mohseni; R.J.H Smith; H. Najmabadi. GJB2 mutation in Baloochi Population. Journal of Genetics, 2008.

Ali Reza Pouya, Seyedeh Sedigheh Abedini, Neda Mansoorian, Valeh Hadavi, Farkhondeh Behjati, Nooshin Nikzat, Sahar Esmaeeli Nieh,Seyed Navid Almadani, Froozan Afroozan, Yousef Shafeghati, Susan Banihashemi, Roxana Kariminejad**, Kimia Kahrizi**, Hossein Najmabadi
Fragile X Syndrome Screening of Families with Consanguineous and Non-consanguineous Parents in the Iranian Population. submitted European Journal of Medical Genetics

Nele Hilgert**, Kimia Kahrizi**, Hossein Najmabadi, Nele Dieltjens, Richard JH Smith, Guy Van Camp. Large deletion in VLGR1 causes Usher syndrome type IIC in male and female patients of an Iranian family. Submitted Am J Med Genet.

Masoud Garshasbi, Valeh Hadavi, Haleh Habibi, **Kimia Kahrizi**, Roxana Kariminejad, Farkhondeh Behjati, Andreas Tzschach,Hossein Najmabadi, Hans Hilger Ropers, Andreas Walter Kuss. A Defect in the TUSC3 Gene is Associated with Autosomal Recessive Mental Retardation. Accepted Am J Hum Genet, Feb 2008

Ramezani, **K. Kahrizi**, M. Razaghiazar, N. Mahdieh and P. Koppens. The frequency of eight common point mutations in CYP21 gene in Iranian patients with congenital adrenal hyperplasia. Iranian Biochemical Journal 12(1): 49-53 2008.

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 Feb;16(2):270-3.

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

M.M. Motazacker; B. Rainer Rost; T. Hucho; M. Garshasbi; **K. Kahrizi**; R. Ullmann; S.S. Abedini; S. Esmaeeli Nieh; S. Hisseini Amini; A. Tzschach; L.R. Jensen; D. Schmitz; H.H. Ropers; H. Najmabadi; A. W. Kuss. A defect in the ionotropic glutamate receptor 6 gene (GRIK2) causes autosomal recessive mental retardation. American Journal of Human Genetics, 81 (4), 792-8, 2007.

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 Jul 15;143(14):1623-9.

K. Kahrizi; E.Keyhani; M. Hasanzad; J.A. Azimian; N. Moradin; B. Shaja Saffar; J. A. Urtizbrea; D. Hantai; H. Najmabadi. Clinical Molecular and Immmunohistochemistry investigation of Neuromuscular disorders in Iranian Populations. Neuro Development Spectrum.27-29 Feb.2007.

L.Van Lear; N. C. Meyer; M. Malekpour; Y. Riazalhosseini; M. Moghanibashi; **K. Kahrizi**; A. Vandevelde; F. Alasti; H. Najmabadi; G.Van Camp; R.J.H. Smith. A Novel DFNA5 mutation does not cause hearing loss in an Iranian family. J. Hum Genet. March 2007.

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, Gruters A, Lenzner S, Jensen LR, Ruschendorf 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 Mar;121(1):43-8. Epub 2006 Nov 21.

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, Gruters A, Lenzner S, Jensen LR, Ruschendorf 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. 2006 Nov 21.

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. 2006 Nov 10.

K. Kahrizi;M. Malekpour; A. Daneshi; H. Najmabadi. Genotype & Phenotype correlation in hearing loss patients of Iranian population. American Journal of Medical Genetics.2006

Marina Kleanthous, Philippos C. Patsalis, Anthi Drousiotou, Mehdi Motazacker, Kyproula Christodoulou, Marios Cariolou, Erol Baysal, **Kimia Khrizi**, Babak Moghimi, Farzin Pourfarzad, Sjozef van Baal, Constantinos Deltas, Hossein Najmabadi, and George P. Patrinos. The Cypriot and Iranian National Mutation Frequency Databases Hum Mutat. 2006 Jun;27(6):598-9.

M. Garshasbi; M. M. Motazacker; **K. Kahrizi**; F. Behjati; S. S. Abedini; S. Esmaeeili Nieh; S. Ghasemi Firouzabadi; Ch. Becker; F. Ruschendorf; P. Nurnberg; A. Tzschach; R. Vazifemand; F. Erdogan; R. Ullmann; S. Lenzner; A. W. Kuss. H. Hiler Ropers; H. Najmabadi. *SNP array-based Homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly.* Hum Genet. Nov 2005 26:1-

8.

Y.Riazalhosseini; Carla nishimura; **K.Kahrizi**; Y. Shafeghati; M.T. Joghataie; M. Mohseni; N. Mahdieh; M. K. Javan; R. H. Smith; N. Najmabadi. DELTA (GJB6-D3S1830) Is Not Common cause of nonsyndromic Hearing loss in the Iranian Population. Arch in Irn Med. 2005 ; 8 (2): 104 – 108.

K. Kahrizi; C. Nishimural; A. Naghavi. Y. Riazalhosseini; R.H.J. Smith; H. Najmabadi. A Novel Mutation of SLC26A4 gene in an Iranian family with Pendred Syndrome. International Journal of Endocrinology & Methabolis. Int J Endocrinol Metab 2005 ; 2 :104-108.

W. Chen, **K. Kahrizi**, NC. Meyer, Y. Riazalhosseini, RF. Mueller, WT. McGuirt, G. Van Camp, H. Najmabadi, RJH. Smith. Mutation of COL11A2 causes Autosomal Ressesive non-syndromic hearing loss at DFNB53 Locus. J Med Genet. 2005 Oct;42(10):e61. Epub 2005 Jul 20.

H. Najmabadi., C. Nishimura., **K. Kahrizi.,** Y. Riazalhosseini., M. Malekpour., A. Daneshi., M. Farhadi., M. Mohseni., N. Mahdieh., A. Ebrahimi, N. Bazazzadegan., A. Naghavi., M. Avenarius., S. Arzhangi., R. J.H. Smith**.** GJB2 Mutation: Passage Through Iran. American Journal of Medical Genetics 9999: 1-6(2005).

L. Abbasi Moheb., M.M. Haghighi., A.R. Ghamari., A. Adibi., H. Rezvan., M. Piontek., S. Hellweg., A. Poorfathollah., **K. Kahrizi**., H. Najmbadi. Expression of Recombinant Human Serum Albumin in the Methylotrophic yeast; Hansenula Polymorpha. The Scientific Journal of Iranian Blood Transfusion Organization- Research Center (2005).

N. Bazazzadegan., N. Mirhosseini., h. Ziaaddini., AR. Asadi., **K. Kahrizi.,** S. Arzhangi., A. Astani., M. Mohseni., Y. Riazalhosseini., N. Mahdieh., Kh. Jalalvand., RJH. Smith., C. Nishimura., H. Najmabadi. Relative frequency of 35delG Mutation in GJB2 in Autosomal Recessive Non-Syndromic Hearing Loss (ARNSHL) Patients in Kerman Population. Journal of Kerman University of Medical Sciences. Summer 2004, Volume 11, Number 3.

B. Moghimi., M. Yavarian., Ch. Oberkanins., S.S.Hosseini Amini., Sh. Khatami., S. Rouhi., **K. Kahrizi.,** H. Najmabadi. Hb Dhonburi (Neapolis) [β126(H4) Val Gly] Identified in a Family fron Northern Iran. Hemoglobin. Vol. 28, No.4, pp.353-356, 2004.

Neishaboury, M., Obercanins, Ch., Abbasi Moheb, L., Pourfathollah, A.A**., Kahrizi, K**., Keyhani, E., Krugluger, W., Najmabadi, H. High prevalence of the –α3.7 Deletion Among Thalassemia Patients in Iran. Hemoglobin, Vol.27, No.1, PP.53-55, 2003.

Najmabadi, H., Neishabury, M., Sahebjam, F., **Kahrizi, K.,** Nikzat, N., Jalalvan, M., Amini, F., Bany Hashemi, S., Moghimi, B., Noorian, A.R., Jannati, A., Mohammadi, M., Javan, M.Kh. The Iranian Human Mutation Gene Bank: A Date and Sample Resource for Worldwide Collaborative Genetic Research. Journal of Human Mutation, Vol 21:146-150(2003.)

Najmabadi, H**.,** Neishabury, M., Sahebjam, F., **Kahrizi, K.**, Nikzat, N., Jalalvan, M., Amini, F., Bany Hashemi, S., Moghimi, B., Noorian, A.R., Jannati, A., Mohammadi, M., Javan, M.Kh. The Iranian Human Mutation Gene Bank: A Date and Sample Resource for Worldwide

Collaborative Genetic Research. Journal of Human Mutation, 2003. 21:146-150

R. A. Cucci., S. Sahebjam., N. Kouchakian., M. Farhadi., **K. Kahrizi.,** S. Arzhangi., N. Daneshmandan., M.Kh. Javan., R. JH. Smith., H. Najmabadi. GJB2 Mutation in Iranian With Autosomal Recessive Non-Syndromic Sensorineural Hearing Loss. Human Mutation, in Brief #504 2002 Online.

H. Njmabadi; Robert A. Cucci; S. Sahebjam; N. Kouchekian; M. Farhadi; **K. Kahrizi;** S. Arzhangi; N. Daneshmandan; Kh. Javan & R. J. H. Smith. GJB2 Mutation in Iranians with Autosomal Recessive non-Syndromic Sensorineural Hearing loss. Human Mutation (2002).

M. Niyshabury; L. Abbasi Moheb; A. Pourfathollah; **K. Kahrizi**; E. Keyhani; W. Krugluger; C. Obercanins; H. Najmabadi. Alpha-Thalassemia Deletion Analysis in Iran. Arch in Irn Med. Oct.2001. 4 (4); 160-164

M. Neishabury., L. Abbasi Moheb., A.A Pourfathollah., **K. Kahrizi.,** E. Keyhani., W. Krugluger., Ch. Obercanins., H. Najmabadi. Alpha Thalassemia in Iran. The American Journal of Human Generics, Oct. 2001.

2. Books

Year	Book title	Publisher	Location
2005	Diagnostic Criteria of Neuromuscular Disorders	USWR	Iran
2004	Duchenne & Becker Muscular Dystrophy	USWR	Iran
2016	Genetics of Aging	USWR	Iran