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Professional profile:

Medical Doctor, Pediatrician, Clinical geneticist fellowship and Professor of Medical Genetics with a significant involvement in designing and conducting studies in the field of medical genetics and writing scientific papers.

Effective collaboration in various national grant in identification of genetic disorders in Iran (Available Documents) and co-director of an approved EU-grant (Gencodys) composed of numbers of European countries and Iran between 2010 and 2015 (Available Documents).

Academic and Professional Appointments:

- University of Social Welfare & Rehabilitation Sciences, Tehran, Iran
  Professor
  Dep: Genetics Research Center
  From 2011 till now
- University of Social Welfare & Rehabilitation Sciences, Tehran, Iran
  Associate Professor
  Dep: Genetics Research Center
  From 2006 to 2011
- University of Social Welfare & Rehabilitation Sciences, Tehran, Iran
  Assistant Professor
  Dep: Genetics Research Center
  From 1999 to 2006

- Welfare Organization, Tehran, Iran
  Deputy for Preventive Affairs
  From 1998 to 1998
- Iran University of Medical Sciences, Tehran, Iran
  Assistant Professor
  Dep: Pediatrics
  From 1994 to 1997
- Iran University of Medical Sciences, Tehran, Iran
  Medical Doctor
  From 1986 to 1994

Areas of Expertise:

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.

Qualifications

1998	Pediatrician. Title: The Causes of Death in Children under 5 years in Tehran Medical
1995	MD. Title: Hypertensive retinopathy

Professional courses

- Genetic Counseling: 10 years, graduate students, University of Social Welfare & Rehabilitation Sciences, Tehran, Iran

- Medical Genetics & Human Genetics: M.S. Genetics students, University of Social Welfare & Rehabilitation Sciences, Tehran, Iran

- Medical Genetics: Ph.D. Medical Genetics students, University of Social Welfare & Rehabilitation Sciences, Tehran, Iran

Books:

1-Diagnostic Criteria of Neuromuscular Disorders. **K. Kahrizi** (2005);

2- Duchenne & Becker Muscular Dystrophy. H. Najmabadi; **K. Kahrizi**; M. Hasanzad; Kh. Javan (2004);

3-Genetics of Aging. T.Akhtar Khavari, H.Najmabadi, **K.Kahrizi** (2016).

Research Supports:

- Genetic investigation of 50 Charcot Marie Tooth patients by Whole Exome Sequencing 2016.

- Genetic investigation of ten Iranian families with autosomal recessive non syndromic hearing loss by homozygosity mapping.

- Clinical, neuroimaging and molecular investigation of congenital Brain malformations in 50 mentally retarded patients associated with microcephaly referred to Genetics Research Center 2015.

- Genetic investigation of five Iranian families with autosomal recessive non syndromic hearing loss by homozygosity mapping.

- Identification of genetic causes of autosomal recessive intellectual disabilitiy associated with ataxia in 15 Iranian families.

- Identification of known genes and loci in syndromic microcephalic Iranian patients (Seckel, Cohen and Rett syndromes).

- Analysis of association serotonin transporter gene and antidepressant response to citaloperam in Iranian major depressive population.

- Screening the children with hearing loss to identify known and unknown genes involved in ARNSHL cases in Iranian population

- A survey on Connexin–26 mutations in patients with genetic deafness in Iranian population.

- Linkage analysis of hearing loss non-syndromic families that they did not map to the follow DNFB1, DFNB2, DFNB3, DFNB4, DFNB9 & DFNB21 loci in our previous study 8 additional loci.

- Investigation of sensorineural autosomal dominant non syndromic hearing loss of known and unknown loci.

- Screening the children with hearing loss to identify known and unknown genes involved in ARNSHL cases in 6 southern provinces of Iran.

- Screening the children with hearing loss to identify known and unknown genes involved in ARNSHL cases in Isfahan province of Iran.

- The Study on inheritance pattern & molecular diagnosis of Connexin 26 mutations in people with hearing impairment covering by Sisatan & Baluchistan Welfare Organization.

- Analysis and detection of fragile X syndrome in children.

- The study of syndromic and non-syndromic mental retardation in Fars province in order to identify known and unknown genes.

- The study of syndromic and non-syndromic mental retardation in Sistan and Baluchistan province in order to identify known and unknown genes.

- The study of relative prevalence of autosomal and X-linked mental retardation patients in four provinces around of Persian Gulf.

- The study of relative prevalence of autosomal and X-linked mental retardation patients in Tehran & Semnan state.

- The study of relative prevalence of autosomal and X-linked mental retardation patients in Gilan state.

- The study of relative prevalence of autosomal and X-linked mental retardation patients in Isfahan state.

- The study of relative prevalence of autosomal and X-linked mental retardation patients of Welfare centers Yazd state.

- The study of relative prevalence of autosomal and X-linked mental retardation patients in X state.

- Analysis of SMA carrier frequency in 100 couples that refer for thalassemia mutation study in country thalassemia screening program to genetic diagnostic laboratory.

- Analysis of spinal muscular atrophy carrier frequency in 200 Iranian healthy blood donors.

- Study on the genes involved in Meniere's disease in Iranian population.
- Analysis of responsively of deaf patients carrying GJB2 mutations after cochlear implantation.
- To determine the prevalence of merosin deficiency in cases of unclassified congenital muscular dystrophy.
- Screening for PDS gene mutations in Pendred syndrome of Iranian population.
- Molecular analysis of autosomal recessive limb girdle muscular dystrophy (LGMD).
- Molecular analysis of autosomal dominant hearing loss.
- Determination of several types of AR LGMD using five common antibodies.
- Molecular analysis of Usher loci in Iranian population.
- Screening of Utrophin in muscle tissue of 15 clinically suspected patients with mild dystrophinopathy (BMD).
- Iranian Human Mutation Gene Bank.
- Immortalization of the DNA bank Cell Line (IHMGB.com).
- Study of prevalence of four known loci in autosomal recessive mental retardation among patients referred to GRC during defined period.
- Investigation of sensorineural autosomal dominant non syndromic hearing loss of 22 known and unknown loci.
- Screening of dystrophin in the muscle tissue of clinically suspected DMD patients with no Xp21 deletion.
- Neuromuscular Classification in Iranian Patients 2005.

Awards:

National Awards:

1. Received the national award as the top 40 researchers by Iranian National Elite Foundation (INSF) and Vice-Presidency for Science and Technology of Iran in 2012 (The first Lady in entire country that selected for this award). (Available document).

2. Received three years research grant provided by Iran National Science Foundation (INSF), 2012-2015 (Available document).

3. Received the national award for distinguished physician award in 2009 by Ministry of Health & Medical Education.

4. Selected as 50 distinguished researcher by Iran Science Elites Federation (ISEF) 2016 and receiving the research grant for one year (Available document).

University Awards:

1. Selected as the distinguished researcher for 8 years in annual awarding of Research Department of University of Social Welfare and Rehabilitation Sciences (Available document).

2. Selected as the distinguished lecturer of medical genetic in annual awarding of Education Department for 10 years (Available document).

Membership:

- American Society of Human Genetics
- European Society of Human Genetics
- Iranian Society of Pediatrics
- Iranian Society of Neurogenetic
- Iranian Society of Human Genetic

Discovery:

Identification a novel syndrome approved by OMIM (Online Mendelian Inheritance in Man) a very well-known and prestigious website for genetic disorders

Publications:

Nikuei P, Kalscheuer VM, Oladnabi M, Wienker TF, Ropers HH, Najmabadi H. Mol Psychiatry. 2019 Jul;24(7):1027-1039

Babanejad M, Zarandy MM, Nikzat N, Bazazzadegan N, Arzhangi S, Mohseni M, **Kahrizi K**, Najmabadi H3. G130V de novo mutation in an Iranian pedigree with nonsyndromic hearing loss without palmoplantar keratoderma. Int J Pediatr Otorhinolaryngol. 2019 Jul 26;126:109607. [Epub ahead of print]

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 Jul 25. doi: 10.1002/humu.23880. [Epub ahead of print]

Beheshtian M, Fattahi Z, Fadaee M, Vazehan R, Jamali P, Parsimehr E, Kamgar M, Faraji Zonooz M, 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 Apr 4. doi: 10.1111/cge.13549. [Epub ahead of print]

InanlooRahatloo K, Peymani F, Kahrizi K, Najmabadi H. Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability Patients. Neuroscience. 2019 Apr 15;404:423-444

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 Feb;117:115-126. Epub 2018 Nov 23.

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. 2018 Dec 14

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

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.

Effect of inbreeding on intellectual disability revisited by trio sequencing. **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. Clin Genet. 2019 Jan;95(1):151-159.

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.

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.

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

Kazeminasab S, Najmabadi H, Kahrizi K. Intellectual Disability and Ataxia: Genetic Collisions. Arch Iran Med. 2018 Jan 1;21(1):29-40. Review. PMID:29664668

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 Jan 12. [Epub ahead of print]

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.

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. Hum Mol Genet. 2017 Dec 23.[Epub ahead of print]

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.

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.

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.

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. GMD2E 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.

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.

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. Mol Psychiatry. 2016 Aug 23. doi: 10.1038/mp.2016.135. [Epub ahead of print]

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. 2016 May 28. doi: 10.1111/cge.12810. [Epub ahead of print]

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.

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