

## Curriculum Vitae

### Personal Information:

**Name:** Afagh

**Surname:** Alavi

**Nationality:** Iranian

**Marital Status:**

**P.O. Box:** 1985713871, Tehran, Iran

**Date of Birth:**

**Tel:** +98-6413224698; +98-916-6421779

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- **2015-now** Assistant Prof. in the University of Social Welfare and Rehabilitation Sciences
- **2014-2015** Post doc researcher in the University of Tehran.
- **2008-2013** PhD student in Cell and Molecular Biology, School of Biology, University College of Science, University of Tehran, Tehran, Iran.

Average of final examination marks: 17.83 (out of 20). Board exam: 16.8(out of 20).

**Title of Thesis:** Linkage analysis in Iranian patients afflicted with Amyotrophic Lateral Sclerosis (ALS) and Autosomal Recessive Congenital Ichthyosis (ARCI)

**Supervisors:** Dr. Elahe Elahi

- **2005-2007** M.Sc. Student in Cell and Molecular Biology, School of Biology, University College of Science, University of Tehran, Tehran, Iran.

Average of final examination marks: 18.81 (out of 20)

**Title of Thesis:** Mutation screening of M1S1 (TACSTD2) gene in Iranian patients afflicted with Gelatinous Drop-like corneal dystrophy (GDLD)

**Supervisors:** Dr. Elahe Elahi

- **1994-1997** B.Sc. Student in Biology, Department of Biology, Shahid Chamran University of Ahvaz, Ahvaz, Iran. Average of final examination marks: 17.75 (out of 20)
- **1990-1993** Bentolhoda High School, Dezfool, Iran. Average of final examination marks: 19.2 (out of 20)

### Teaching Experience

Teacher of Biology at high Schools in several cities in Iran, **Sept. 1997-2013.**

Teacher of Biology for other teachers in Tehran (Chardangeh), 2011

Teacher of genetics at the College Of Environment; Karaj, 2012-2013

Teacher of human genetics, cancer genetics, genetic engineering, advanced molecular genetics, advanced genetic engineering, advanced cancer genetics, bioinformatics and genetics of mitochondria at the University of University of Social Welfare and Rehabilitation Sciences; Tehran, 2014-now

## Research Experience

- Genetic analysis of Iranian patients affected to Gelatinous Drop-like corneal dystrophy, Granular, Avellino and Lattice corneal dystrophies.
- Genetic analysis of Iranian patients affected to Juvenile open-angle glaucoma (JOAG).
- Hybridization of multiplex PrASE products to oligonucleotide spotted microarrays (Microarray in Juvenile open-angle glaucoma (JOAG) patients).
- Genetic analysis of Iranian patients affected to Autosomal recessive Congenital Ichthyosis patients.
- Linkage analysis in Iranian patients affected to Amyotrophic Lateral Sclerosis (ALS), Autosomal Recessive Congenital Ichthyosis (ARCI), neurodegeneration with brain iron accumulation (NBIA), Neimann-Pick C disease patients, ....
- Exome sequencing of Iranian Amyotrophic Lateral Sclerosis (ALS), Neimann-Pick C, Parkinson disease, hyperinsulinemic hypoglycemia, neuropathies, and Hereditary Spastic Paraplegia (HSP) patients.
- Repeat-primed PCR for hexanucleotide expansions in *C9orf72* gene among Iranian ALS and PD patients.
- DNA barcoding in fish
- Southern blotting for Facioscapulohumeral muscular dystrophy (FSHD) patients
- Genetic study of muscular dystrophies; limb girdle muscular dystrophies, Sarcoglycanopathies ...

## Workshop and Training

- Participation in an advanced course on “Genetic Association Studies”, taught by Heather J. Cordell (Institute of Human Genetics, Newcastle University, UK) at the Research Center for Gastroenterology and Liver Diseases, Shaheed Beheshti University of Medical Sciences, Tehran, Iran, Jan. 24-25 2007.
- Participation in an advanced course on Neurological disease. Imam hospital, Tehran, Iran, 1390/9/2. Tehran, Iran, 1390/9/2.
- Participation in Applications of FISH technique workshop. University of Social welfare and Rehabilitation Sciences. 1398.
- Participation in Array CGH workshop. University of Social welfare and Rehabilitation Sciences. 1398.
- Participation in CRISPR/Cas workshop. University of Social welfare and Rehabilitation Sciences. 1398.
- Participation in Tertiary for exome sequencing workshop. University of Social welfare and Rehabilitation Sciences. 1398.

- Participation in RNA-sequencing workshop. University of Social welfare and Rehabilitation Sciences. 1398.
- Participation in Drosophila melanogaster workshop. University of Social welfare and Rehabilitation Sciences. 1398.

## Publications

### Articles:

1. **Alavi A**, Elahi E, Tehrani MH, Amoli FA, Javadi MA, Rafati N, Chiani M, Banihosseini SS, Bayat B, Kalhor R, Amini SS. Four mutations (three novels, one founder) in *TACSTD2* among Iranian GDLD patients. *Invest Ophthalmol Vis Sci*. 2007; 48(10):4490-7.
2. **Alavi A**, Elahi E, Amoli FA, Tehrani MH. Exclusion of *TACSTD2* in an Iranian GDLD pedigree. *Mol Vis*. 2007; 13:1441-5.
3. **Alavi A**, Elahi E, Rahmati-Kamel M, Karimian F, Rezaei-Kanavi M. Mutation Screening of *TGFBI* in Two Iranian Avellino Corneal Dystrophy Pedigrees. *Clin Experiment Ophthalmol*. 2008; 36(1):26-30.
4. Bayat B, Yazdani SH, **Alavi A**, Chiani M, Chitsazian F, Khoramian Tusi B, Suri F, Narooie-Nejhad M, Sanati MH, Riazuddin S, Elahi E. Contributions of *MYOC* and *CYP1B1* mutations to JOAG. *Mol Vis*. 2008; 14:508-17.
5. **Alavi A**, Mirshams Shahshahani M, Elahi E. Manifestation of diffuse yellowish keratoderma on the palms and soles in autosomal recessive congenital ichthyosis patients may be indicative of mutations in *NIPAL4*. *J Dermatol*. 2012; 39(4):375-81.
6. **Alavi A**, Nafissi S, Rohani M, Zamani B, Sedighi B, Shamschiri H, Fan JB, Ronaghi M, Elahi E. Genetic analysis and SOD1 mutation screening in Iranian amyotrophic lateral sclerosis patients. *Neurobiol Aging*. 2013; 34(5):1516.e1-8.
7. **Ansari Dezfouli M**, **Alavi A**, Rohani M, Rezvani M, Nekuie T, Klotzle B, Tonekaboni SH, Shahidi GA, Elahi E (**\* These two authors contributed equally to this manuscript**). *PANK2* and *C19orf12* mutations are common causes of neurodegeneration with brain iron accumulation. *Mov Disord*. 2013; 28(2):228-32.
8. Ansari Dezfouli M, Jaberi E, **Alavi A**, Rezvani M, Shahidi GA, Elahi E, Rohani M, Pantothenate kinase 2 mutation with eye-of-the-tiger sign on magnetic resonance imaging in three siblings. *Ir J neurol*. 2012; 11(4): 1-4.

9. **Alavi A**, Nafissi S, Shamshiri H, Malakooti Nejad M, Elahi E. Identification of mutation in *NPC2* by exome sequencing results in diagnosis of Niemann-Pick disease type C. *Mol Genet Metab*. 2013; 110(1-2): 139–144.
10. **Alavi A**, Nafissi S, Rohani M, Shahidi GA, Zamani B, Shamshiri H, Safari I, Elahi E. Repeat expansion in *C9ORF72* is not a major cause of ALS among Iranian patients. *Neurobiol Aging*. 2014; 35(1): 267.e1e267.e7
11. **Alavi A**, Khani M, Nafissi S, Shamshiri H, Elahi E. An Iranian FALS patient with p.Val48Phe mutation in exon 2 of the *SOD1* gene: a clinical and genetic report. *The Iranian Journal of Basic Medical Sciences (Iran J Basic Med Sci)*, Vol. 17, No. 10, Oct 2014
12. **Alavi A**, Shamshiri H, Nafissi S, Khani M, Klotzle B, Fan J, Steemers F, Elahi E. HMSN-P caused by p.Pro285Leu mutation in TFG is not confined to patients with Far East ancestry. *Neurobiol Aging*. 2015; 36: 1710-1716
13. Khani M, **Alavi A**, Nafissi S, Elahi E. Observation of p.Asn86Ser causing mutation in *SOD1* in an Iranian ALS patient and evidencing absence of genotype/phenotype correlation for this mutation. *Iran J Neurol* 2015; 14(3).
14. Khani M, Shamshiri H, **Alavi A**, Nafissi S, Elahi E. Identification of novel TFG mutation in HMSN-P pedigree: Emphasis on variable clinical presentations. *J Neurol Sci*. 2016; 369:318-23. doi: 10.1016/j.jns.2016.08.035
15. **Alavi A**, Malakouti Nejad M, Shahidi G, Elahi E. Mutations in *C19orf12* and intronic repeat expansions in *C9orf72* not observed in Iranian Parkinson's disease patients. *Neurobiol Aging*. 2017; 54: 214.e11-214.e12. DOI: 10.1016/j.neurobiolaging.2017.03.020.
16. Rohani M, Shahidi G, **Alavi A**, Lang A, Yousefi N, Razme S, Fasano A. Tremor-dominant pantothenate kinase-associated neurodegeneration. *Movement Disorders Clinical Practice*. 2017;4(5): 772-774. DOI: 10.1002/mdc3.12512.
17. **Alavi A\***, Esmaeili S, Nilipour Y, Nafissi S, Tonekaboni SH, Zamani G, Ashrafi MR, Kahrizi K, Najmabadi H, Jazayeri F. LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. *J Neurogenet*. 2017; 31(3):161-169. DOI: 10.1080/01677063.2017.1346093.
18. Rohani M, Lang AE, Sina F, Elahi E, Fasano A, Hardy J, Bras J, **Alavi A\***. Action myoclonus and

seizure in Kufor-Rakeb syndrome. *Movement Disorders Clinical Practice*. 2018; 5(2): 195-199. DOI: 10.1002/mdc3.12570.

19. **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 Apr;28(4):303-314. doi: 10.1016/j.nmd.2018.01.001.

20. Nozari A, Aghaei-Moghadam E, Zeinaloo A, Mollazadeh R, Majnoon M, **Alavi A**, Ghasemi Firouzabadi S, Mohammadzadeh A, Banihashemi S, Nikzaban M, Najmabadi H, Behjati F. A novel splicing variant in FLNC gene responsible for a highly penetrant familial dilated cardiomyopathy in an extended Iranian family. *Gene* 659 (2018) 160–167.

21. Nozari A, Aghaei-Moghadam E, Zeinaloo A, **Alavi A**, Ghasemi Firouzabadi S, Minaee S, Eskandari Hesari M, Behjati F. A Pathogenic Homozygous Mutation in Pleckstrin Homology Domain of *RASA1* Gene Responsible for Familial Tricuspid Atresia in an Iranian Consanguineous Family. *Cell Journal (Yakhteh)* Volume 21, Number 1, Spring 2019 (Apr-Jun), Serial Number: 81.

22. Rohani M, Fasano A, Lang AE, Javanparast L, RahimiBidgoli MM, **Alavi A\***. Pantothenate Kinase Associated Neurodegeneration mimicking Tourette Syndrome. *Neurol Sci*. 2018 Jun 21. doi: 10.1007/s10072-018-3472-5.

23. Khani M; **Alavi A**; Shamshiri H; Zamani B; Hassanpour H; Kazemi MM; Nafissi S; Elahi E. Mutation screening of SLC52A3, C19orf12, and TARDBP in Iranian ALS patients. *Neurobiol Aging*. 75:225.e9-225.e14. doi: 10.1016/j.neurobiolaging.2018.11.003.

24. Rohani M, Fasano A, Haji Akhondi F, Haeri G, Lang AE, RahimiBidgoli MM, Javanparast L, Zamani B, Shahidi G, **Alavi A\***. Beta-propeller protein associated neurodegeneration (BPAN); the first report of three patients from Iran with de novo novel mutations. *Parkinsonism and Related Disorders*. 2018 Nov 13. pii: S1353-8020(18)30496-6. doi: 10.1016/j.parkreldis.2018.11.012.

25. Rahmani B, Fekrmandi F, Ahadi K, Ahadi T, **Alavi A**, Ahmadiani A, Asadi S. A novel nonsense mutation in WNK1/HSN2 associated with sensory neuropathy and limb destruction in four siblings of a large Iranian pedigree. *BMC Neurol*. 2018 Nov 29;18(1):195. doi: 10.1186/s12883-018-1201-6.

26. Zare-Abdollahi D, Bushehri A, **Alavi A**, Dehghani A, Mousavi-Mirkala M, Effati J, Miratashi SAM, Dehani M, Jamali P, Khorram Khorshid HR. MFSD8 gene mutations; evidence for phenotypic

heterogeneity. *Ophthalmic Genet.* 2019 Apr;40(2):141-145. doi: 10.1080/13816810.2019.

27. Khani M, Taheri H, Shamshiri H, Houlden H, Efthymiou S, **Alavi A**, Nafissi S, Elahi E. Continuum of phenotypes in hereditary motor and sensory neuropathy with proximal predominance and Charcot–Marie–Tooth patients with TFG mutation. *Am J Med Genet.*2019;1–9.

28. Khani M, Shamshiri H, Fatehi F, Rohani M, Haghi Ashtiani B, Haji Akhoundi F, **Alavi A**, Moazzeni HR, Taheri H, Tolou Ghani M, Javanparast L, Hashemi SS, Haji Seyed Javadi R, Heidari M, Nafissi S, Elahi E. Problematic differential diagnosis of ARHSP and JALS in some families with SPG11 mutations. *Mol Genet Genomic Med.* 2020 Jul;8(7):e1240. doi: 10.1002/mgg3.1240.

29. Hajati R, Rahimi Bidgoli MM, Rohani M, **Alavi A\***. An Overview of Neurodegeneration with Brain Iron Accumulation (NBIA) syndromes and the disease status in Iranian population. *Tehran University Medical Journal.* 2020;78(2);58-68.

30. **Alavi A**, Mokhtari M, Hajati R, Davarzani A, Fasano A, Lang AE, Rohani M. Late-Onset Mitochondrial Membrane Protein-Associated Neurodegeneration With Extensive Brain Iron Deposition. *Mov Disord Clin Pract.* 2019 Nov 30;7(1):120-121. doi: 10.1002/mdc3.12868.

31. Pashaei M, Rahimi Bidgoli MM, Zare-Abdollahi D, Najmabadi H, Haji-Seyed-Javadi R, Fatehi F, **Alavi A\***. The second mutation of *SYCE1* gene associated with autosomal recessive non-obstructive azoospermia. *J Assist Reprod Genet.* 2020 Feb;37(2):451-458. doi: 10.1007/s10815-019-01660-1.

32. **Alavi A**, Darki F, Bidgoli MMR, Zare-Abdollahi D, Moini A, Shahshahani MM, Fischer J, Elahi E. Mutation in *ALOX12B* likely cause of POI and also ichthyosis in a large Iranian pedigree. *Mol Genet Genomics.* 2020 Jul;295(4):1039-1053. doi: 10.1007/s00438-020-01663-z.

33. Rahimi Bidgoli MM, Javanparast L, Rohani M, Najmabadi H, Zamani B, **Alavi A\***. CAPN1 and hereditary spastic paraplegia: a novel variant in an Iranian family and overview of the genotype-phenotype correlation. *Int J Neurosci.* 2020 May 13:1-13. doi: 10.1080/00207454.2020.1763344.

34. Bushehri A, Zare-Abdollahi D, **Alavi A**, Dehghani A, Mousavimikala M, Khorram Khorshid HR. Identification of *PROS1* as a Novel Candidate Gene for Juvenile Retinitis Pigmentosa. *Int J Mol Cell Med.* 2019 Summer;8(3):179-190. doi: 10.22088/IJMCM.BUMS.8.3.179.

35. Fattahi M, Bushehri A, **Alavi A**, Asghariazar V, Nozari A, Ghasemi Firouzabadi S, Motamedian Dehkordi P, Javid M, Farajzadeh Valiliou S, Karimian J, Behjati F. Bi-allelic Mutations in *ALDH5A1* is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability. *Gene.* 2020 Jul 1:144918. doi: 10.1016/j.gene.2020.144918.

36. Farajzadeh Valilou S, **Alavi A**, Pashaei M, Ghasemi Firouzabadi S, Shafeghati Y, Nozari A, Hadipour F, Hadipour Z, Maghsoodlou Estrabadi B, Gholamreza Noorazar S, Banihashemi S, Karimian J, Fattahi M, Behjati F. Whole-Exome Sequencing Identifies Three Candidate Homozygous Variants in a Consanguineous Iranian Family with Autism Spectrum Disorder and Skeletal Problems. *Mol Syndromol*. 2020 Jun;11(2):62-72. doi: 10.1159/000506530. Epub 2020 Mar 11.
37. Haeri G, Akhoundi FH, **Alavi A**, Abdi S, Rohani M. Endocrine Abnormalities in a Case of Neurodegeneration with Brain Iron Accumulation. *Mov Disord Clin Pract*. 2020 Jun 24;7(6):706-707. doi: 10.1002/mdc3.12990. eCollection 2020 Aug.
38. Kuźma-Kozakiewicz M, Andersen PM, Elahi E, **Alavi A**, Sapp PC, Morita M, Żekanowski C, Berdyński M. Putative founder effect in the Polish, Iranian and United States populations for the L144S SOD1 mutation associated with slowly uniform phenotype of amyotrophic lateral sclerosis. *Amyotroph Lateral Scler Frontotemporal Degener*. 2020 Aug 10:1-6. doi: 10.1080/21678421.2020.1803359.
39. Khani M, Taheri H, Shamshiri H, Moazzeni H, Hardy J, Bras JT, InanlooRahatloo K, **Alavi A**, Nafissi S, Elahi E. Deep geno- and phenotyping in two consanguineous families with CMT2 reveals HADHA as an unusual disease-causing gene and an intronic variant in GDAP1 as an unusual mutation. *J Neurol*. 2020 Sep 8. doi: 10.1007/s00415-020-10171-4.
40. Vafae-Shahi M, Ghasemi S, Ghahvechi Akbar M, Tahernia L, Davarzani A, Hajati R, Zare-Abdollahi D, **Alavi A\***. Giant axonal neuropathy: The first Iranian case with a variation in the gigaxonin gene and a glance to the other cases. *Current Journal of Neurology*. Accepted.

#### Conference Abstracts (Poster presentations):

1. **Alavi A**, Javadi M, Rafati N, Bayat B, Banoei MM, Elahi E. Novel missense mutation in *M1S1* gene in Iranian Gelatinous Drop-Like Corneal Dystrophy (GDLD) patients. European Human Genetics Conference, Amsterdam-The Netherlands, May 6–9, 2006.
2. **Alavi A**, Hosseini Tehrani M, Asadi Amoli F, Elahi E. Y184C Missense mutation in *TACSTD2* in an Iranian Gelatinous Drop-like Corneal Dystrophy (GDLD) pedigree. The 2nd International Congress of Biochemistry and Molecular Biology, Shiraz-Iran, Oct 29-Nov 1, 2007.
3. Nezari H, Kalhor R, Banihosseini SS, Suri F, **Alavi A**, Zargar SJ, Ahmadian A, Elahi E. Hybridization of multiplex PrASE products to oligonucleotide spotted microarrays. The 2nd International Congress of Biochemistry and Molecular Biology, Shiraz-Iran, Oct 29-Nov 1, 2007.
4. Banihosseini S, Kazemi MH, Shojaee SM, **Alavi A**, Shahidi G, Sina F, Zamani B, Sadeghi H, Parsa K, Elahi E. Mutation screening of exon 3 of Parkin gene in a young cohort of Iranian PD patients. European Human Genetics Conference, Barcelona-Spain, May 31–June 3, 2008.

5. **Alavi A**, Heidary Arash E, Inanloo K, Banihosseini S, Elahi E. Estimation of *CFTR* mutation carrier frequency based on known frequency of p.F508del in Iranian neonates. European Human Genetics Conference, Gothenburg-Sweden, June 12–15, 2010.
6. **Alavi A**, Mirshams Shahshahani M, Moazzeni HR, Elahi E. Homozygosity mapping in a large Iranian pedigree affected with Autosomal Recessive Congenital Ichthyosis (ARCI) reveals linkage to region encompassing *NIPAL4*/Ichthyn. European Human Genetics Conference, Amsterdam-The Netherlands, June 12–15, 2011.
7. Suri F, Zargar SJ, Yazdani S, **Alavi A**, Elahi E. One genotype-six different phenotype: variable expression not incomplete penetrance. 15 National & 3rd International Conference of Biology, Tehran-Iran, August 19-21, 2008.
8. Inanloo K, Elahi E, **Alavi A**, Jaberi E. Mutation Screening of *MEF2A* Gene in Iranian afflicted with Coronary Artery Disease. The First International Student Congress on Cell and Molecular Medicine, Shiraz-Iran. February 17-19, 2011.
9. **Alavi A**, Mirshams Shahshahani M, Malakooti Nejad M, Rasooli P, Elahi E. Homozygosity mapping in one Iranian pedigree affected with Autosomal Recessive Congenital Ichthyosis (ARCI) reveals linkage to region 17p13 and mutation in *ALOX12B* gene. European Human Genetics Conference, Nürnberg-Germany, June 23-26, 2012.
10. **Alavi A**, Mirshams Shahshahani M, Safari I, Elahi E. Homozygosity mapping in 2 autosomal recessive congenital ichthyosis pedigrees reveals linkage to regions 5q33 and 17p13. 2012; 17<sup>th</sup> Iranian and 5<sup>th</sup> international Biology congress, Kerman-Iran, September 4-6, 2012.
11. Malakouti Nejad M, **Alavi A**, Hashemi M, Shahidi GA, Elahi E. Mutation screening of *ATP13A2* in early onset Iranian Parkinson's disease patients. European Human Genetics Conference, Nürnberg-Germany, June 23-26, 2012.
12. **Alavi A**, Elahi E, Nafissi S, Shamshiri H, Houshmand M, Malakooti Nejad M. Exome sequencing revealed a *NPC2* mutation in an Iranian Niemann Pick C-type 2 disease family. European Human Genetics Conference, Paris-France, June 8-11, 2013.
13. **Alavi A**, Elahi E, Malakooti Nejad M, Homozygosity mapping in an Iranian pedigree affected with muscular dystrophy limb girdle (LGMD) reveals linkage to 2p12-14 and 10q25-26 chromosomes. 3rd International Student Biotechnology Congress, Tehran-Iran, May 6-8, 2013.
14. Malakouti Nejad M, Elahi E, Jaberi E, **Alavi A**, Hashemi M, Shahidi G. disease caused by a novel *ATP13A2* truncating mutation. European Human Genetics Conference, Paris-France, June 8-11, 2013.
15. **Alavi A**, Elahi E, Khani M, Malakooti Nejad M. Homozygosity mapping in an Iranian pedigree affected with muscular dystrophy limb girdle (LGMD) reveals linkage to chromosome 2p12-14 and a mutation in Dysferlin gene. European Human Genetics Conference, Milan-Italy, May 31- June 3, 2014.



16. Malakouti Nejad M, Elahi E, **Alavi A**, Shahidi G. Repeat expansion in C9ORF72 is not a common cause of Parkinson's disease among Iranian patients. European Human Genetics Conference, Milan-Italy, May 31- June 3, 2014.
17. **Alavi A**, Jaberi E, Elahi E, Nafissi S, Shamshiri H, Rohani M. Clinical application of whole exome sequencing in diagnosis of undiagnosed inherited diseases. First international & 13<sup>th</sup> Iranian genetics congress, Tehran-Iran, May 24-26, 2014. **Best presentation Poster in first international & 13th Iranian genetics congress**
18. Khani M, Elahi E, **Alavi A**, Nafissi S, Shamshiri H. TARDBP mutations are not a common cause of amyotrophic lateral sclerosis in Iranian patients. First international & 13<sup>th</sup> Iranian genetics congress, Tehran-Iran, May 24-26, 2014.
19. Suri F, **Alavi A**, Elahi E, Shamshiri H. Homozygosity mapping used for identification of disease causing genes in heterogenic disorders. First international & 13<sup>th</sup> Iranian genetics congress, Tehran-Iran, May 24-26, 2014.
20. Homozygosity mapping in an Iranian pedigree affected with early onset Parkinson's disease (EOPD) and linkage to chromosome 6. 2nd International Conference on Parkinson's Disease & Movement Disorders. USA-Chicago, November 28-30, 2016.
21. Khani M, Elahi E, **Alavi A**, Nafissi S, Malakooti Nejad M. Screening of TARDBP in Iranian amyotrophic lateral sclerosis (ALS) patients. European Human Genetics Conference, Milan-Italy, May 31- June 3, 2014.
22. **Alavi A**, Elahi E, Shamshiri H, Nafissi S, Moghadam A. HMSN-P is a new type of neuronopathy caused by mutation in TFG gene. European Human Genetics Conference, Glasgow, Scotland-United Kingdom, June 6-9, 2015.
23. **Alavi A**, Elahi E, Shamshiri H, Nafissi S. HMSN-P: is a new type of neuronopathy caused by mutation in TFG gene; is not confined to patients with Far East ancestry. The 5th International Congress on Neuropathic Pain (NeuPSIG). France-Nice, May 14-17, 2015.
24. Malakouti Nejad M, Elahi E, **Alavi A**, Shahidi G. Screening for C9ORF72 repeat expansions among Iranian patients with Parkinson's disease. First international & 13<sup>th</sup> Iranian genetics congress, Tehran-Iran, May 24-26, 2014.
25. Esmaeili S, **Alavi A\***, Nafissi S. Phenotypic presentations of Iranian Patients affected to Facioscapulohumeral muscular dystrophy (FSHD). Second International & Fourteenth Iranian Genetics Congress. Tehran-Iran, May 21-23, 2016.
26. **Alavi A**, Elahi E, Malakooti Nejad M, Shahidi GA. Homozygosity mapping in an Iranian pedigree affected with early onset Parkinson's disease (EOPD) and linkage to chromosome 6. 2th international conference on Parkinson's disease and movement disorders. Phoenix, USA, Dec 05-07, 2016.

27. Khani M, Elahi E, Shamshiri H, **Alavi A**, Nafissi S. P.Gly269Val mutation in TFG identified as cause of disease in second Iranian HMSN-P pedigree. 5<sup>th</sup> Basic and clinical neuroscience congress, Tehran-Iran, December 7-9, 2016.
28. **Alavi A**, Esmaeili S, ..... FSHD. 3rd International Conference on Parkinson's Disease & Movement Disorders. USA-Chicago, September 25-26, 2017.
29. Javanparast L, Rahimi Bidgoli MM, **Alavi A**, Rohani M. Clinical features and results of genetic analysis of three Iranian patients affected to pantothenate kinase associated neurodegeneration (PKAN). 6<sup>th</sup> Basic and clinical neuroscience congress, Tehran-Iran, December 20-22, 2017.
30. **Alavi A**, Ghorbanpour E , Pourmajidian M, Elahi E,d, Shahidi G. Linkage to a new locus on chromosome 6 in an Iranian pedigree diagnosed with early onset Parkinson's disease (EOPD). 2nd International Conference on Chronic Diseases. Berlin, Germany, June 25-26, 2018.
31. Ghorbanpour E, Rahimi Bigoli MM, Javanparast L, **Alavi A**, Rohani M. Genotype and phenotype analysis of two unrelated patients with Beta-Propeller protein-Associated Neurodegeneration (BPAN). 5<sup>th</sup> International Conference on Parkinson's Disease & Movement Disorders. Baltimore, Maryland, USA, October 19-20, 2018.
32. **Alavi A**, Pashaei M, Rahimi Bidgoli MM, Javan Parast L, Rohani M, Fatehi F, Nafissi S, Kahrizi K, Najmabadi H. Hereditary spastic paraplegia (HSP) is more heterogeneous than it appears. European Conference of Human Genetics. Milan, Italy June 16-19 2018.
33. Javan Parast L, **Alavi A**, Rahimi Bidgoli MM, Pashaei M, Rohani M, Fatehi F, Nafissi S, Kahrizi K, Najmabadi H. Identification of causative genes (three known and one novel) in four unrelated Iranian families affected to hereditary spastic paraplegia (HSP) using whole exome sequencing. 3<sup>rd</sup> international and 15<sup>th</sup> national Iranian genetics congress. Tehran, Iran. May 13-15, 2018.
34. Fattahi M, **Alavi A**, Ghasemi Firouzabadi S, NozariA, Farajzadeh Valilou S, Karimian J, Crosby A, Behjati F. Whole exome sequencing identified a novel ALDH5A1 variant associated with SSADH Deficiency in an Iranian family with autism. 3<sup>rd</sup> international and 15<sup>th</sup> national Iranian genetics congress. Tehran, Iran. May 13-15, 2018.
35. Javadi Golrodbari F, **Alavi A**, Kazemi Nasab S, Kahrizi K, Najmabadi H. Identification of novel genes in ten patients with Neuromuscular disease who have no mutations in known genes by reanalyzing data of whole exome sequencing. 3<sup>rd</sup> international and 15<sup>th</sup> national Iranian genetics congress. Tehran, Iran. May 13-15, 2018.
36. Rahimi Bidgoli MM, **Alavi A\***, Javan Parast L, Pashaei M, Fatehi F. Exome sequencing reveals *SYCE1* mutation associated with autosomal recessive azoospermia in an Iranian family; the second report in the worldwide. 8<sup>TH</sup> Yazd international congress & student

award in Reproductive medicine & third congress of reproductive genetics. Yazd, Iran. April 19-21, 2019.

37. Rahimi Bidgoli MM, **Alavi A\***, Rohani M, Javan Parast L, Pashaei M, Fatehi F, Nafissi S, Kahrizi K, Najmabadi H. Genetics heterogeneity and novel genes in autosomal recessive Hereditary Spastic Paraplegia (AR-HSP). European Human Genetics Conference, Gothenburg, Sweden. June 15–18, 2019.
38. Davarzani A, Hajati-Kenarsari R, Zare-Abdollahi D, Vafaei Shahi M, **Alavi A\***. Clinical and genetic analysis of an Iranian patient affected with Giant axonal neuropathy. 20<sup>th</sup> Annual Research congress of Iranian Medical Sciences Students. Kermanshah, Iran, 5-8 Sahrivar 1398.
39. Hajati-Kenarsari R, Davarzani A, Rohani M, **Alavi A\***. Mitochondrial Membrane Protein Associated Neurodegeneration (MPAN) with an extensive and atypical pattern of brain iron deposition. 20<sup>th</sup> Annual Research congress of Iranian Medical Sciences Students. Kermanshah, Iran, 5-8 Sahrivar 1398.
40. Hajati, A. Davarzani, S. Hashemi, M. Rahimi Bidgoli, L. Javan Parast, M. Pashaei, F. DanaeeFard, H. Najmabadi, M. Rohani, F. Fatehi, S. Nafissi, **A. Alavi\***. Genetic analysis of 50 unrelated cases affected with hereditary spastic paraplegia (HSP): a novel candidate gene. European Human Genetics Conference (Virtual), Vienna, Austria. June 6-9, 2020.
41. Hajati R, Davarzani A, Rohani M, InanlooRahatloo K, **Alavi A\***. Woodhouse-Sakati Syndrome: The first case report from Iran. 21th annual research congress of Iranian medical sciences Students, Babol, Iran. 21-24 Mordad 1399.
42. Davarzani A, Hajati R, Rohani M, InanlooRahatloo K, **Alavi A\***. Charcot-Marie-Tooth (CMT) type 2A2A with pyramidal signs due to a mutation in the MFN2 gene: A Case Report. 21th annual research congress of Iranian medical sciences Students, Babol, Iran. 21-24 Mordad 1399.
43. Hashemi SS, Zare-Abdollahi D, InanlooRahatloo D, DanaeeFard F, Rohani M, **Alavi A\***. Three families affected to primary COQ10 deficiency: Importance of whole exome sequencing in diagnosis of this group of disorders. 21th annual research congress of Iranian medical sciences Students, Babol, Iran. 21-24 Mordad 1399.

#### **Full articles presented at conferences (Oral presentations):**

1. **Alavi A**, Javadi MA, Rafati N, Bayat B, Banoei MM, Elahi E. Two novel putative disease causing mutations in *M1S1* gene in Iranian Gelatinous Drop-like Corneal Dystrophy (GDLD) patients. 9th Iranian Genetics Congress, Tehran-Iran, May 20-22, 2006.

2. **Alavi A**, Elahi E, Mirshams Shahshahani M, Inanloo K, Moazzeni HR. Mutation in Ichthyn/*NIPAL4* is the cause of Autosomal recessive Congenital Ichthyosis in a large Iranian family with yellowish palmoplantar keratoderma. The first International Student Congress on Cell and Molecular Medicine (ISCCMM), Shiraz-Iran, February 17-19, 2011.  
**Best presentation award in 1st International Congress on Cell and Molecular Medicine**
3. **Alavi A**, Mirshams Shahshahani M, Malakooti Nejad M, Elahi E. Homozygosity mapping in two large Iranian pedigrees affected with autosomal recessive congenital ichthyosis (ARCI) reveals linkage to regions 5q33 and 17p13. 2011. The 1th international and 5th Annual Congress of Iranian Neurogenetic Society, Tehran-Iran, November 23-25, 2011.
4. Malakouti Nejad M, **Alavi A**, Hashemi M, Shahidi GA, Elahi E. Novel *ATP13A2* mutation associated with early onset Parkinson's disease. Basic and clinical neuroscience congress, Tehran-Iran, November 7-9, 2012.
5. **Alavi A**, Elahi E, Nafissi S, Roohani M, Zamani B, Sedighi B, Shamshiri H, Malakooti Nejad M. Genetic analysis and *SOD1* mutation screening in Iranian amyotrophic lateral sclerosis patients (ALS). The 6th Annual Congress of Iranian Neurogenetic Society. Kerman-Iran, December 8-13, 2012.
6. Rohani M, **Alavi A**, Zamani B, Elahi E. Familial ALS. The second Iranian congress of neuromuscular disorders and electrodiagnosis. Tehran-Iran, 4-8 July, 2012.
7. Elahi E, Nafissi S, Rohani M, **Alavi A**, Khani M, Shamshiri H. Genetics of ALS in Iran. First international & 13<sup>th</sup> Iranian genetics congress, Tehran-Iran, May 24-26, 2014.
8. **Alavi A**, Elahi E, Nafissi S, Rohani M, Khani M, Shamshiri H. Frequency of *SOD1*, *C9orf72*, and *TARDBP* mutations in Iranian ALS patients. 3<sup>rd</sup> Basic and clinical neuroscience congress, Tehran-Iran, October 29-31, 2014.
9. **Alavi A**, Elahi E, Malakooti Nejad M, Shahidi G. Homozygosity mapping in an Iranian pedigree affected with early onset Parkinson's disease (EOPD) and linkage to chromosome 6. The 8th Annual Congress of Iranian Neurogenetic Society, Ahvaz-Iran, January 21-23, 2015.
10. Elahi E, **Alavi A**. Amyotrophic lateral sclerosis (ALS). The 12<sup>th</sup> congress & workshop of rare diseases. Tehran-Iran, 26 Feb- 2 March, 2014.
11. Elahi E, **Alavi A**, Shamshiri H, Khani M, Nafissi S. Iranian patients affected with familial forms of neuromuscular disorders with mutations in *TFG*. 57<sup>th</sup> annual meeting of the Japanese society of neurology. Japan-Kobe, May 18-21, 2016.
12. Yousefi N, **Alavi A**, Safari I, Elahi E. Obesity associated locus sought by linkage analysis and exome sequencing in an extended pedigree. 23<sup>rd</sup> international student congress of (Bio) medical sciences. Netherlands- Groningen, 7-10, 2016.

13. Esmaeili S, **Alavi A\***, Nafissi S. Genetic diagnosis of facioscapulohumeral muscular dystrophy (FSHD) using southern blotting with non-radioactive probes. 1 national conference on new findings in biology. Iran-Zahedan, Nov 23-24, 2016.
14. Esmaeili S, **Alavi A\***, Kahrizi K, Najmabadi H, Nafissi S. Genetic analysis of 20 FacioScapuloHumeral muscular Dystrophy (FSHD) families using southern blotting with non-radioactive probes. 3th international conference on molecular medicine. Iran-Isfahan, Dec 15-16, 2016.
15. **Alavi A\***. LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. سمینارهای یکروزه ژنتیک پزشکی تشخیصی- تحقیقی- دانشکده پیراپزشکی دانشگاه علوم پزشکی شهید بهشتی Sep 15, 2017.
16. Khani M, Elahi E, Nafissi S, **Alavi A**, Shamshiri H, Taheri H, Tolou Ghani M, Moazzeni H. Identification of causative genes for the neurodegenerative diseases ALS, BVVL, Fazio Londe, CMT2 and HMSN-P. 3<sup>rd</sup> International & 15<sup>th</sup> Iranian Genetics Congress. May 13-15 2018.
17. Behjati F, Ghasemi Firouzabadi S, kariminejad R, Vameghi R, Banihashemi S, Jamali P, Firouzkouhi Moghaddam M, Farbod Mofidi Tehrani H, Dehghani H, Karimian J, Farajzadeh Valilou S, Fattahi Nafchi M, Pashaei M, **Alavi A**, Crosby A, Najmabadi H. Copy Number Variation (CNV) analysis and Next generation Sequencing (NGS), in some Iranian Patients with Autism. 3<sup>rd</sup> International & 15<sup>th</sup> Iranian Genetics Congress. May 13-15 2018.
18. **Alavi A\***. Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients. سمینارهای یکروزه ژنتیک پزشکی تشخیصی- تحقیقی- بیمارستان صارم June 22, 2018.
19. **Alavi A**, Rahimi Bidgoli MM, Rohani M, Javan Parast L, Pashaei M, Fatehi F. Genetics heterogeneity of Hereditary Spastic Paraplegia (HSP) is more than it appears. سمینارهای یکروزه ژنتیک پزشکی تشخیصی- تحقیقی- بیمارستان صارم Jan 25, 2019.
44. Rahimi Bidgoli MM, **Alavi A\***, Javan Parast L, Pashaei M, Rohani M, Nafissi S, Kahrizi K, Najmabadi H. Diagnostic rates of whole exome sequencing in Hereditary Spastic Paraplegia. 20<sup>th</sup> Annual Research congress of Iranian Medical Sciences Students. Kermanshah, Iran, 5-8 Sahrivar 1398.
20. Hashemi SS, Zare-Abdollahi D, InanlooRahatloo K, DanaeeFard F, Rohani M, **Alavi A\***. Application of whole exome sequencing (WES) in clinical diagnosis; An example. International congress of Isfahan Biomedical Sciences. Sept. 26- Oct.1, 2020.

**Contributor in research grant**

1. Mutation screening of *M1S1* gene in Iranian Gelatinous Drop-Like corneal Dystrophy patients.
2. Linkage analysis in Iranian families affected with autosomal recessive congenital ichthyosis (ARCI). Pasteur institute
3. Linkage analysis in Iranian patients affected with Amyotrophic Lateral Sclerosis (ALS). INSF
4. Analysis of hexanucleotide expansions in *C9orf72* gene among Iranian patients affected with Amyotrophic Lateral Sclerosis (ALS).
5. Identification of causative gene for an atypical neuromuscular disease with motor and sensory manifestations using linkage analysis and exome sequencing. INSF
6. Genetic analysis of 10 Iranian patients affected to FSHD (Facioscapulohumeral muscular dystrophy). GRC-USWR
7. Clinical and genetic study of 40 Iranian patients affected to Limb girdle muscular dystrophy type sarcoglycopathy. INSF
8. Finding of causative gene in an Iranian family affected to primary ovarian failure (POF) using whole exome sequencing (WES) and screening of candidate gene in other Iranian POF patients. BMN
9. Identification of disease causative gene in a large Iranian pedigree affected to primary ovarian failure (POF). INSF
10. Searching for causative genes in 10 Iranian patients affected to hereditary spastic paraplegia (HSP) using whole exome sequencing. USWR
11. Searching for causative genes in 50 Iranian patients affected to hereditary spastic paraplegia (HSP) using whole exome sequencing. GRC-USWR-NIMAD

## **Research Interests**

- Human Genetics
- Genetic analysis of neurodegenerative diseases including, Amyotrophic Lateral Sclerosis (ALS), hereditary spastic paraplegia (HSP), Neurodegeneration with brain Iron accumulation (NBIA), Parkinson Disease, Neuropathies, .... and muscular dystrophies.
- Genetic analysis of primary ovarian failure (POF)
- Gene finding approaches (SNP genotyping, whole exome sequencing, ....)