Aida Ghasemi

Specialty: Human Genetics

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Research Links:

- ResearchGate Profile: https://www.researchgate.net/profile/Aida-Ghasemi
- Google Scholar Profile: https://scholar.google.com/citations?hl=en&user=RvJRs3UAAAAJ

Education

Date	Degree	Duration	Institution	Country/City	Major/Situation	Average mark
2020-2023	M.Sc.	3 years	University of social welfare and rehabilitation (USWR)	Iran/ Tehran	Human Genetics *Talented student*	19.05 / 20
2016-2020	B.Sc.	4 years	Shiraz university of medical sciences (SUMS)	Iran/ Shiraz	Medical Laboratory Sciences *Talented student*	18.15 / 20
2012-2016	Diploma	4 years	Farzanegan high school (SAMPAD)	Iran/ Abadeh	Experimental Science	19.90 / 20

Research Interests

- Neurodegenerative & Neuromuscular disorders (e.g., Amyotrophic Lateral Sclerosis (ALS), Hereditary Spastic Paraplegia (HSP), Neurodegeneration with Brain Iron Accumulation (NBIA), Neuroacanthocytosis (NA), Charcot Marie Tooth (CMT), Mucolipidosis, Myasthenic Syndromes, Primary Familial Brain Calcification (PFBC), Myopathy))
- Gene finding approaches (e.g., whole exome sequencing)
- Human/ Medical Genetics

Fields of Specialization

Laboratory Skills: General & Medical laboratory techniques (Sampling, Chemistry, Hematology, Microbiology, Immunology, Cytology)

Genetics Techniques: Human/Medical Genetics (DNA extraction, RNA extraction, PCR, real-time PCR, NGS analyzing (secondary & tertiary), Primer design, Cell culture, Gel extraction, Electrophoresis, Karyotyping, Sanger sequencing, etc.)

Scientific Investigation: Particularly in neurodegenerative and neuromuscular diseases

Writing and Editing: Translating, writing, and revising scientific articles

Software Proficiency: Microsoft office, Ubuntu, IGV, GeneRunner, Codoncode aligner, Sequencher, Automap, Annovar, Expansion hunter, BWA, Endnote, SPSS, and various bioinformatics software

Language Ability

Persian: Native

English: Fluent (Diploma from Iranian language institute)

M.Sc. Thesis Title

Evaluation of copy-number variations (CNVs) in 15 Iranian hereditary spastic paraplegia (HSP)-affected families to find the probably disease-causing genetic variants.

Thesis score: 20 / 20

Publication

Journals

Year	Journal name	Article title	Vol. & Page
2022	Neurological Sciences	The second family affected with a *PRDM8*-related disease	2022 Jun;43(6):3847-3855. doi: 10.1007/s10072-021-05815-w.
2023	Razi Journal of Medical Sciences	An overview of amyotrophic lateral sclerosis (ALS) and its genetic status	RJMS 2023; 30 (1)
2023	Neuromuscular Disorders	Expanding the phenotypic spectrum of *NMNAT1* variants	2023 Feb 8;33(4):295-301. doi: 10.1016/j.nmd.2023.02.001.
2023	Molecular Syndromology	Description of phenotypic heterogeneity in a *GJC2*-related family and literature review	2023 Oct;14(5):405-415. doi: 10.1159/000529678.
2023	Molecular Syndromology	Copy number variations (CNVs) in hereditary spastic paraplegia (HSP)-related genes; Evaluation of an Iranian HSP cohort and literature review	2023 Dec;14(6):477-484. doi: 10.1159/000531507.

2023	Neurological Sciences	The first reports of *FA2H*-associated neurodegeneration from two unrelated Iranian families	2023 Dec;44(12):4359-4362. doi: 10.1007/s10072-023-06932-4.
2024	Neurological Sciences	*JAM2* variants can be more common in Primary familial brain calcification (PFBC) cases than those appear; May be due to a founder mutation	2024 Aug;45(8):3829-3844. doi: 10.1007/s10072-024-07419-6.
2024	Orphanet Journal of Rare Diseases	*COLQ*-Congenital Myasthenic Syndrome in an Iranian Cohort: The Clinical and Genetics Spectrum	2024 Mar 12;19(1):113. doi: 10.1186/s13023-024-03116-x.
2024	Neuromuscular Disorders	*CHRNE*-related Congenital Myasthenic Syndrome in Iran: Clinical and Molecular Insights	2024 Oct 25:46:105234. doi: 10.1016/j.nmd.2024.105234.
2024	Neurogenetics	Clinical and Genetic Diversity in Iranian Individuals with *RAPSN* -related Congenital Myasthenic Syndrome	2024 Nov 26;26(1):9. doi: 10.1007/s10048-024-00787-3.
2024	Neurogenetics	Three rare subtypes of hereditary spastic paraplegia (HSP): SPG76, SPG56, and SPG69	2024 Nov 28;26(1):12. doi: 10.1007/ s10048-024-00787-1.
2024	Molecular Genetics and Genomics	Beginning at the ends: Telomere and telomere- based cancer therapeutics	2024 Dec 6;300(1):1. doi: 10.1007/s00438-024-02206-6.
2024	Pediatric Neurology	Genetic homogeneity of a *TDP1* variant, c.1478A>G, as the disease-causing variant of Spinocerebellar ataxia with axonal neuropathy 1 (SCAN1) in Middle East and a Systematic Review	2024 Dec 30:164:41-52. doi: 10.1016/j.pediatrneurol.2024.12.011.
2025	Canadian Journal of Neurological Sciences	A novel homozygous variant in the *MCOLN1* gene associated with severe oromandibular dystonia	2025 Jan;52(1):110-118. doi: 10.1017/cjn.2024.47.
2025	Neuropathology	Phenotypic and Genotyping spectrum of two Iranian cases with *RBCK1*-associated polyglucosan body myopathy	2025 Feb;45(1):48-54. doi: 10.1111/neup.12993.
2025	Neuromuscular Disorders	Identifying novel *AGRN* variants in congenital myasthenic syndrome; Insights from three Iranian families	2025 Apr;49:105342. doi: 10.1016/j.nmd.2025.105342.
2025	Neurological Sciences	Mutation Spectrum and Clinical Features of *MYORG* in Iranian Patients with Primary Familial Brain Calcification	2025 Mar 22. doi: 10.1007/s10072-025-08105-x.

2025	Journal of the Peripheral Nervous System	*GDAP1*-related Charcot-Marie-Tooth disease: axonal or demyelinating subtype? autosomal recessive or autosomal dominant inheritance?	2025 Sep;30(3):e70046. doi: 10.1111/jns.70046.
2025	Current Genetic Medicine Reports	Myo-Neuropathy and Congenital Bilateral Cataract Due to a *GFER* Variant in an Iranian Family	Accepted
2025	Neuromuscular Disorders	Clinical and molecular spectrum of an Iranian Charcot-Marie-Tooth 2T cohort, and the diagnostic importance of Whole Exome Sequencing in identifying co-occurrence inherited disorders	Under review
2025	Bratislava Medical Journal	Charcot-Marie-Tooth Type 4C Mimicking CIDP: Diagnostic Pitfalls and Genetic Confirmation	Under review
2025	Muscle and Nerve	A Cohort of Iranian Patients with Congenital Myasthenic Syndrome due to Glycosylation Defects	Under review

Books

Year	Book title	Publisher	Location
2022	Translating Emery's Elements of Medical Genetics and Genomics 2022	Rouyan Pajooh	Iran/ Tehran

Research Paper Presented in Congresses / Seminars / Symposia

Poster presentations

Year	Congress/Symposium	Paper title	Location
May 2021	9th Yazd International Congress & Student Award in Reproductive Medicine with 4th Reproductive Genetics	The role of the *SYCE1* gene in the male and female fertility; A literature review	Iran/ Yazd
December 2021	International Conference on Human Genetics and Genomics	The important role of copy-number variations in hereditary spastic paraplegia	Iran/ Yazd
December 2021	10th Basic and Clinical Neuroscience Congress	Genetic heterogeneity of hereditary spastic paraplegia (HSP) in Iranian population	Iran/ Tehran
March 2022	8th National & 2nd International Webinar on Medical Genetics: Diagnostics & Research	SPG4 and SPG11 are the most common types of hereditary spastic paraplegia (HSP) in Iran	Iran/ Mashhad

June 2022	European Human Genetics Conference (EHGC)	Genetics heterogeneity and novel genes in a cohort of Iranian patients with hereditary spastic paraplegia (HSP)	Austria/ Vienna
June 2024	European Human Genetics Conference (EHGC)	A founder mutation in the *COQ7* gene, may result in a pure form of Hereditary Spastic Paraplegia (HSP)	Germany/ Berlin
May 2024	31st Congress of Neurology and Clinical Electrophysiology of Iran	Autosomal recessive than autosomal dominant forms of FAHR syndrome are more severe as well as, more prevalent in the Iranian population	Iran/ Tehran
May 2024	6th International and 18th Iranian Genetics Congress	Advantage of whole exome sequencing (WES) for identification of disease-causing variant in less known genes for hereditary spastic paraplegia (HSP)	Iran/ Tehran
November 2024	8th International Congress on Biomedicine	Identification of novel and known variants in the *JAM2* gene in the Iranian families affected to Primary familial brain calcification (PFBC)	Iran/ Tehran
May 2025	European Human Genetics Conference (EHGC)	The *TDP1*:c.1478A>G variant as the main cause of Spinocerebellar ataxia with axonal neuropathy 1 (SCAN1): a possible founder mutation in the Middle East	Italy/ Milan
September 2025	6th Iranian Congress of Neuromuscular and Electrodiagnostic Medicine	Clinical and Genetic Characterization of five Iranian families affected to triple A syndrome or Allgrove syndrome with neurological dysfunction	Iran/ Tehran
September 2025	6th Iranian Congress of Neuromuscular and Electrodiagnostic Medicine	Expanding the Phenotypic Spectrum of *GFER* Variants: Myopathy, Neuropathy, and Congenital Cataracts in an Iranian Family	Iran/ Tehran

September 2025	6th Iranian Congress of Neuromuscular and Electrodiagnostic Medicine	Unveiling Demyelinating Features in an Iranian Patient with *MTRFR* Mutation	Iran/ Tehran
Oral presentations			
September 2022	23rd Medical Students Annual National Research Congress & the 9th Medical Students International Research Congress	Application of whole-exome sequencing to detect of copy number variations (CNVs) in hereditary spastic paraplegia (HSP)	Iran/ Ardabil
February 2023	4th Research Congress of Hormozgan Medical Science Students	Identification of *MCOLN1* and *C19orf12* gene variants in two Iranian families with brain iron accumulation	Iran/ Hormozgan
March 2023	5th International and 17th Iranian Genetic Congress	Evaluation of whole exome sequencing (WES) efficiency in the genetic diagnosis of 85 Iranian families affected with hereditary spastic paraplegia (HSP)	Iran/ Tehran
April 2023	1st International Symposium of Walking on DNA Toward Clinics	Intrafamilial clinical heterogeneity in an Iranian multi-affected family with *GJC2* gene mutation	Iran/ Ahvaz
May 2023	1st Annual Student Congress of Medical Sciences Universities in Tehran	Identification of two novel variants in the *MYORG* gene in the Iranian families affected to Primary familial brain calcification (PFBC)	Iran/ Tehran
May 2023	1st Annual Student Congress of Medical Sciences Universities in Tehran	The advantage of whole-exome sequencing for the detection of disease-causing variants in patients with hereditary spastic paraplegia	Iran/ Tehran

Research Grant

Title	Grant number	Position
Searching for the disease-causing variant in a family affected with a type of neurodegenerative disorder with brain iron accumulation, using whole exome sequencing (WES) technique.	2957	Principal investigator
Identification of causative genes in 10 Iranian families affected to hereditary motor and sensory neuropathy using whole exome sequencing.	68456	Principal investigator

Searching for causative genes in five Iranian patients affected to idiopathic calcification of basal ganglia (FAHR) using whole exome sequencing (WES).	2888	Co-Principal investigator
Genetic study of the second group of Iranian patients with idiopathic basal ganglia calcification (FAHR).	3030	Co-Principal investigator
Searching for causative genes in 15 Iranian patients affected to hereditary spastic paraplegia (HSP) using whole exome sequencing (WES).	2592	Co-Principal investigator
Searching for causative genes in 15 Iranian patients affected to hereditary spastic paraplegia (HSP) using whole exome sequencing (WES).	1969	Co-Principal investigator
Investigation the symptoms and para-clinical features of *RAPSN* mutation in patients suffering from neurology clinic from 1390 to 1402.	69654	Co-Principal investigator
Evaluating of transcriptome and gene expression alteration in a family with hereditary spastic paraplegia (HSP) using RNA-sequencing method.	3231	Co-Principal investigator
Evaluation of genotype-phenotype correlation in patients with congenital myasthenic syndrome with *CHRNE* gene mutation.	73662	Co-Principal investigator
Investigation of the clinical and electrophysiological features, and genetic variants in Iranian patients with congenital myasthenic syndrome due to mutations in the *AGRN* gene	79665	Co-Principal investigator
Investigation and analysis of the relationship between clinical symptoms and molecular features in at least 12 Iranian patients with polyneuropathy caused by mutations in the *GDAP1* gene.	79679	Co-Principal investigator
Study of the genotype-phenotype correlation in 10 Iranian families with Charcot-Marie-Tooth disease due to mutations in the *MME* gene and analysis of 6 families with shared mutations	79752	Co-Principal investigator
An assessment of the diagnostic yield from the reanalysis of whole exome sequencing data in ten Iranian patients suffering from hereditary neuropathy who have not reached a genetic diagnosis	92729	Co-Principal investigator

Reviewing Experience (manuscript/abstract)

Year	Journal	Publisher
2025	European Journal of Pediatrics	Springer Nature
2025	Current Genetic Medicine Reports	Springer Nature
2025	Bratislava Medical Journal	Springer Nature
2025	Acta Neurologica Belgica	Springer Nature
2025	BMC Medical Genomics	Springer Nature

Work Experience

Year	Duration	Location
2022	1 month	Endocrine & Metabolism Research Institute, Tehran university of medical sciences, Tehran, Iran
2023	1 month	Azin Genetics Laboratory, Tehran, Iran
2023-until now	-	Neuromuscular Research Center, Tehran University of Medical Sciences, Tehran, Iran

Teaching Experience

Year	Position	Duration	Institution/Course	Location
April 2021	Lecturer	2 hours	Laboratory tests	Shahid Sadoughi University of Medical Sciences and Health Services, Yazd, Iran
May 2022	Lecturer	10 hours	14th National Medical Students Scientific Olympiad	Shahid Sadoughi University of Medical Sciences and Health Services, Yazd, Iran
Jan 2019 - Jan 2021	Mentor	2 years	Mentoring Program	Shiraz University of Medical Sciences and Health Services, Shiraz, Iran

Awards and Fellowships

Year	Congresses / Symposia	Award
2022	European Human Genetics Conference (EHGC)	Conference fellowship
2022	23rd medical students annual national research congress & the 9th medical students international research congress	Best presenter
2023	4th Research Congress of Hormozgan medical science students	Best presenter
2023	1st international symposium of walking on DNA toward clinics	Best key talk
2023	1st annual student congress of medical sciences universities in Tehran	Best presenter