Curriculum Vitae

Hassan Saei, M.Sc. Ph.D.

Imagine Institute of Genetic Diseases
Laboratory of Hereditary Kidney Diseases
University Paris Cite, Paris, France

Contacts & Profiles

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Education

Ph.D. | Pasteur Paris Universite | Paris, France **2021 - 2024**
Genetics. Imagine Institute of Genetic Diseases (PPU Fellowship)
Laboratory of Hereditary Kidney Diseases – Directed by Prof. Corinne Antignac

M.Sc. | Iran University of Medical Sciences | Tehran, Iran **2017 - 2020**
Human genetics.

B.Sc. | University of Tabriz | Tabriz, Iran **2013 - 2017**
Biology

Research Experience

Postdoctoral Researcher | Imagine Institute of Genetic Diseases, Paris, France (Jan 2025)
Research Focus: WIDGeT consortium project – Developing reprogramed viral vectors (AAV) for the targeted treatment of podocytopathies
Main Projects:

- Developing in vitro binding and library screening assays
- Performing computational analysis of AAV transduction in different models

Ph.D. Candidate | Imagine Institute of Genetic Diseases, Paris, France (Oct 2021-Dec 2024)
Research Focus: Hereditary kidney disease genetic diagnosis and model design for drug discovery
Main Projects:

- Developing a computational pipeline for genetic diagnosis of ADTKD-*MUC1* using short-read sequencing data
- Identification and validation of missing variants in X-linked Alport Syndrome (XLAS)
- Development of XLAS kidney organoid model for variant characterization and drug discovery
- Development of XLAS in vivo model with deep-intronic variation for testing therapeutic approaches

Pasteur Institute | Paris, France.

Main PhD training and certificates:

- Scientific Integrity (EMBO), Bioinformatics program for PhD students: R and Statistics track, Bioimage analysis track (Fiji and super-resolution Images).
- Bioinformatics track: Unix, Metagenomics, Single cell data analysis, Functional analysis, Proteomics data analysis, Utilities for HTS data analysis, Expression quantification and differential analysis, Variant calling, ChIP-seq data analysis (87 hours)
- Laboratory animal experimentation – Designer diploma (59 hours)
- Oral presentation workshop (4 hours)
- Journal clubs (24 hours)

M.Sc. | Iran University of Medical Sciences | Tehran, Iran

Research Focus: Rare disease genetic diagnosis with NGS, Functional characterization of complex disease

Main Project:

- Whole-exome sequencing for genetic diagnosis of patients with Maple syrup urine disease (MSUD)
- Bioinformatic analysis via in house pipelines for variant detection, classification and in silico analysis of the identified variations
- Modeling the pre-eclampsia disease using placenta cell lines for studying disease mechanism

Research assistant | Shahid Akbar-Abadi Obstetrics and Gynecology Hospital, Tehran, Iran

Mentor: Maryam Abiri, Ph.D.

Main duties:

- Grant writing with my supervisor: NIMAD 2019 grant winner
- Workshop lecturer on next-generation sequencing data analysis in USERN, medical genetics network, Tehran

Teaching Assistant | Ali-Asghar Children Hospital | Iran University of Medical Science, Tehran

Mentor: Saeed Talebi, M.D., Ph.D.

Main duties:

- Analyzing patient's whole exome sequencing raw data from Fastq to clinical report.
- Seminars for master student on high-throughput sequencing data analysis.

Analytic and Computational Skills

Molecular biology:

Skilled in a range of molecular genetic techniques, including nucleic acid extraction, protein isolation, Sanger sequencing, cell culture, RNAi transfection (ASOs), Western blotting, SDS-PAGE, cloning, 2D

and 3D immunofluorescence (IF), as well as imaging techniques such as Spinning disk confocal microscopy (SDCM).

Bioinformatics:

Proficient in UNIX command line and bash scripting, utilizing R for comprehensive data analysis (bulk/single-cell RNAseq) and visualization, and Python for both package development and further data analysis and visualization tasks. Experienced with High-performance computing (HPCs), adept at working with containers, and proficient in building apptainer/docker images.

Sequence analysis:

Proficient in a variety of bioinformatic tools and software for tasks such as sequence analysis (Sequencher, Alamut, SpliceAI), variant detection (GATK, deepVariant), annotation (CADD, SIFT, MutationTaster), visualization (IGV), interpretation (in silico variant effect prediction), and population databases (GnomAD).

Other:

Capable of quickly learning and adapting to new computational tools for biological data analysis. Willing to take the advantages of machine learning models and algorithms in solving biological problems.

Peer-reviewed Publications

1. **Saei H**, Estebe B, Gaudin N, Esmailpour M, Haure J, Gribouval O, Arrondel C, Moriniere V, Tian P, Lennon R, Antignac C, Mollet G, Dorval G. Therapeutic splice modulation of COL4A5 reinstates collagen IV assembly in an organoid model of X-linked Alport syndrome. **Under revision in *JCI insight***. 2025.[Link]
2. Kachmar J*, **Saei H***, Morinière V, Heidet L, Knebelmann B, Gribouval O, Mautret-Godefroy M, Burtey S, Vuiblet V, Alla A, Ibalanky A, Moranne O, Nizon M, Savenkoff B, Nitschké P, Antignac C, Doral G. Phenotypic heterogeneity of ADTKD-MUC1 diagnosed using VNtyper, a novel genetic technique. AJKD. 2025. **(*co-first authors**) [Link]
3. **Saei H**, Masson C, Morinière V, Kachmar J, Heidet L, Gribouval O, Antignac C, Dorval G. Using VNtyper from whole exome sequencing data to detect pathogenic variants in the MUC1 gene. **JASN**. 2024. [Link]
4. Boisson M, Arrondel C, Cagnard N, Morinière V, Arkoub ZA, **Saei H**, Heidet L, Kachmar J, Hummel A, Knebelmann B, Bonnet-Dupeyron MN, Isidor B, Izzedine H, Legrand E, Couarch P, Gribouval O, Bole-Feysot C, Parisot M, Nitschké P, Antignac C, Dorval G. A wave of deep intronic mutations in X-linked Alport syndrome. **Kidney Int**. 2023 Aug; 104(2):367-377. [Link]

5. **Saei H**, Morinière V, Heidet L, Gribouval O, Lebbah S, Tores F, Mautret-Godefroy M, Knebelmann B, Burtey S, Vuiblet V, Antignac C, Nitschké P, Dorval G. VNtyper enables accurate alignment-free genotyping of *MUC1* coding VNTR using short-read sequencing data in autosomal dominant tubulointerstitial kidney disease. **iScience**. 2023 Jun 17; 26(7):107171. [Link]
6. Abiri M*, **Saei H***, Eghbali M, Karamzadeh R, Shirzadeh T, Sharifi Z, Zeinali S. Maple syrup urine disease mutation spectrum in a cohort of 40 consanguineous patients and insilico analysis of novel mutations. **Metab Brain Dis**. 2019 Aug; 34(4):1145-1156. **(*co-first authors**) [Link]
7. **Saei H**, Govahi A, Abiri A, Eghbali M, Abiri M. Comprehensive transcriptome mining identified the gene expression signature and differentially regulated pathways of the late-onset preeclampsia. **Pregnancy Hypertens**. 2021 Aug; 25:91-102. [Link]
8. Eghbali M, Fatemi KS, Salehpour S, Abiri M, **Saei H**, Talebi S, Olyaei NA, Yassaee VR, Modarressi MH. Whole-Exome Sequencing Uncovers Novel Causative Variants and Additional Findings in Three Patients Affected by Glycogen Storage Disease Type VI and Fanconi-Bickel Syndrome. **Front Genet**. 2021 Jan 11; 11:601566. [Link]
9. Jabbarpour N*, **Saei H***, Jabbarpoor Bonyadi MH, Bonyadi M. Identification of novel cis-mutations in the *GJA8* gene in a 3-generation Iranian family with autosomal dominant congenital nuclear cataract. **Ophthalmic Genet**. 2022 Oct; 43(5):609-614. **(*co-first authors)** [Link]

Memberships and Conference Proceedings

- The American College of Medical Genetics and Genomics (ACMG) | Member 2023-Present
- The American Society of Human Genetics (ASHG) | Member 2023-Present
- The European Society of Human Genetics (ESHG) | Member
- Poster presentation at NephGen Symposium 2023 in Freiburg, Germany (*Poster Prize winner)
- Oral presentation in the Podocyte meeting in Philadelphia, PA, USA, 2023 (*Travel award winner, Presented online)
- Poster presentation in ESHG 2018 conference, Milan, Italy.

Awards and honors

- Pasteur-Paris University International Ph.D. Fellowship, France (PPU-Imagine, 2021)
- Poster Prize winner, NephGen Symposium, Freiburg, Germany 2023.
- Travel award winner, 14th International Podocyte Conference, PA, USA, 2023.
- Silver medal and 2nd place winner in national Biology Olympiad for undergraduate students, Ministry of Higher Education, Iran
- Outstanding student award in basic science at University of Tabriz from Ministry of Higher Education, Iran.

- Iran National Elite's foundation conference travel award, 2018.
- 1st class honour in the B.Sc. amongst 31 students, University of Tabriz, Tabriz, Iran.

References

Corinne Antignac, M.D, Ph.D. (Laboratory director)

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Guillaume Dorval, M.D., Ph.D. (Ph.D. co-supervisor)
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Saeed Talebi, M.D., Ph.D. (Human genetics program director)
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All Degrees and Certificates are available on request