

Hassan Saei, MSc. PhD.

Imagine Institute of Genetic Diseases
Laboratory of Hereditary Kidney Diseases (U1163), 75015 PARIS
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EDUCATION

Université Paris Cité, Institut Pasteur, France

2021-2024

Ph.D. in Genetics

Bio Sorbonne Paris Cite, DGNRV department

Fellowship from PPU-Imagine Program

Iran University of Medical Sciences, Iran

2017-2020

M.Sc. in Human Genetics

Overall GPA: 4/4

University of Tabriz, Iran

2013-2017

B.Sc. in Biology

Overall GPA: 3.84/4

*All credentials are evaluated by World Education Services [Link]

PROFESSIONAL EXPERIENCES

Imagine Institute of Genetic Diseases, Paris, France

Jan 2025 - Present

Postdoctoral Researcher

Paris, France

- Research Focus: AAV targeted therapy development for podocytopathies in partnership with Sanofi, and White-Lab Genomics
- Developing in vitro and in vivo binding and transduction screening assays and their computational analysis

Oct 2021 - Dec 2024

Ph.D. Candidate

Paris, France

- Research Focus: Improving genetic diagnosis in hereditary renal diseases and development of robust models for therapy development
- Developed a computational pipeline for variant detection in the coding-VNTR in ADTKD-MUC1 using short-read sequencing data (vntyper.org)
- Integrated genomic and transcriptomic analyses to enhance the genetic diagnosis of X-linked Alport syndrome
- Developed a splice-modulation therapeutic approach for Alport syndrome using human kidney organoids

Iran University of Medical Sciences

Oct 2018 - Sep 2020

M.Sc. Internships

Tehran, Iran

- Research Focus: Rare disease genetic diagnosis with NGS, Functional characterization of complex disease
- Exome sequencing for diagnosis of patients with MSUD
- Bioinformatic analysis via in house pipelines for variant detection, classification and in silico analysis of the new variants
- Modeling pre-eclampsia disease using 2D culture

Oct 2018 - Dec 2019

Research assistant

Tehran, Iran

- Shahid Akbar-Abadi Obstetrics and Gynecology Hospital. Mentor: Dr. Maryam Abiri, Ph.D.
- Grant writing with my supervisor: NIMAD 2019 grant winner to study pre-eclampsia

- Ali-Asghar Children Hospital. Mentor: Dr. Saeed Talebi, M.D., Ph.D.
- Analyzing exome sequencing from raw data to clinical report
- Workshop lecturer on next-generation sequencing data analysis in USERN, medical genetics network, Tehran

ANALYTIC AND COMPUTATIONAL SKILLS

Programing Languages	Python, R/Markdown, Bash, High Performance Computing, Git
Workflow Management	Docker, Singularity
Big Data Analysis	RNA-seq, scRNA-seq, scATAC-seq, Proteomics
Sequence analysis	GATK, deepVariant, CADD, SIFT, MutationTaster, IGV
Stem cell research	hiPSC Maintanance and Differentiation
Disease models	Kidney organoid, Murine model characterization

MAIN TRAINING AND CERTIFICATES

Pasteur Institute

Oct 2021 - Dec 2024

PhD courses

Paris, France

- Scientific Integrity (EMBO)
- Bioinformatics program for PhD students: R and Statistics track, Bio-image analysis track
- 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
- Laboratory animal experimentation – Designer Diploma

AWARDS, SCHOLARSHIPS AND FUNDINGS

- 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, National Biology Olympiad for undergraduate students, Ministry of Higher Education, Iran
- Outstanding Student Award in Basic Science, University of Tabriz, Ministry of Higher Education, Iran
- Iran National Elite's Foundation Conference Travel Award, 2018
- 1st class honour in the B.Sc. (ranked 1/31 students), University of Tabriz, Iran

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. Splice modulation of COL4A5 reinstates collagen IV assembly in an organoid model of Alport syndrome. *JCI Insight*. 2025. [Full text]
2. Petzold, F, Jeanpierre C, Chen X, Morinière V, Benmerah A, Dorval G, **Saei H**, Heidet L, Antignac C, Saunier S. Exome Sequencing in a Large Cohort with Ciliopathy-Related Kidney Disease. *CJASN*. 2025. [Full text]
3. 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, Dorval G. Phenotypic Heterogeneity of ADTKD-MUC1 Diagnosed Using VNtyper, a Novel Genetic Technique *American Journal of Kidney Diseases*. 2025. (* co-first authors) [Full text]

4. **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. [Full text]
5. 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. [Full text]
6. **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. [Full text]
7. 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 in silico analysis of novel mutations. *Metab Brain Dis*. 2019 Aug; 34(4):1145-1156. (*co-first authors) [Full text]
8. **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. [Full text]
9. 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. [Full text]
10. Jabbarpour N*, **Saei H***, Jabbarpour 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)

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, Freiburg, Germany, 2023 (*Poster Prize winner)
- Oral presentation at the Podocyte Meeting, Philadelphia, PA, USA, 2023 (*Travel Award winner, presented online)
- Poster presentation at ESHG Conference, Milan, Italy, 2018

REFERENCES

- **Corinne Antignac, M.D., Ph.D.**
Professor and Director
Laboratory of Hereditary Kidney Diseases, INSERM U1163, Imagine Institute, Paris, France
Email: Corinne.antignac@inserm.fr
- **Geraldine Mollet, Ph.D.**
Associate Professor, HDR (Ph.D. Supervisor)
Laboratory of Hereditary Kidney Diseases, INSERM U1163, Imagine Institute, Paris, France
Email: Geraldine.mollet@inserm.fr

- **Guillaume Dorval, M.D., Ph.D.**

Assistant Professor (Ph.D. Co-supervisor)

Laboratory of Hereditary Kidney Diseases, INSERM U1163, Imagine Institute, Paris, France

Genetics Department, Faculty of Medicine, University Paris Cité, France

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- **Saeed Talebi, M.D., Ph.D.**

Associate Professor (Human Genetics Program Director)

Medical Genetics Department, Faculty of Medicine, Iran University of Medical Sciences, Tehran, Iran

Email: Talebi.s@iums.ac.ir