Hassan Saei, MSc. PhD.

Imagine Institute of Genetic Diseases
Laboratory of Hereditary Kidney Diseases (U1163), 75015 PARIS
Email — Personal website — GitHub — LinkedIn

PROFESSIONAL PROFILE

Postdoctoral researcher in human genetics with strong expertise in both wet- and dry-lab research. Experienced in kidney organoid disease modeling, RNA-based therapeutics, and AAV-mediated gene therapy for inherited nephropathies. Early in my Ph.D., I developed a bioinformatics tool that significantly improved the genetic diagnosis of patients with *ADTKD-MUC1* using short-read sequencing data. I subsequently focused on developing physiologically relevant *in vitro* kidney organoid models to evaluate therapeutic approaches. My research integrates genomics, computational analysis, and functional validation to accelerate translational discovery and precision medicine strategies.

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 (WIDGeT consortium project)
- 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 of hereditary renal diseases and developing robust disease models for therapeutic discovery
- Developed a splice-modulation therapeutic approach for Alport syndrome using human kidney organoids
- Developed a computational pipeline for variant detection in the coding-VNTR in ADTKD-MUC1 using short-read sequencing data (vntyper-online)
- Integrated genomic and transcriptomic analyses to enhance the genetic diagnosis of X-linked Alport syndrome

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

- Modeling pre-eclampsia using 2D culture, biomarker identification and their functional analysis
- 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

Oct 2018 - Dec 2019

Research assistant *Tehran, Iran*

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

Oct 2017 - Dec 2018

Teaching Assistant *Tehran, Iran*

- 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

Programming 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	Human iPSC culture and differentiation
Disease models	Kidney organoid development, Mouse model characterization

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**[*], 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)

MAIN TRAINING AND CERTIFICATES

Pasteur Institute Oct 2021 - Dec 2024
PhD courses *Paris, France*

- Scientific Integrity (EMBO)
- Laboratory animal experimentation – Designer Diploma
- 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

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

MEMBERSHIPS AND CONFERENCE PROCEEDINGS

- The American College of Medical Genetics and Genomics (ACMG) — Member, 2023–2024
- The American Society of Human Genetics (ASHG) — Member, 2023–2024
- The European Society of Human Genetics (ESHG) — Member
- Poster presentation at ESHG Conference, Milan, Italy, 2025
- Poster presentation BaCell3D. Basel, Switzerland, 2025
- Poster presentation at American Society of Matrix Biology, basement membrane workshop, Manchester, UK, 2024 (Travel award winner)

- Poster presentation at ESHG Conference, Berlin, Germany, 2024
- Oral presentation European Renal Association, Stockholm, Sweden, 2024.
- 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
  Email: guillaume.dorval@inserm.fr, guillaume.dorval@aphp.fr
- **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
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