# Hassan Saei, MSc. PhD.

Imagine Institute of Genetic Diseases

Laboratory of Hereditary Kidney Diseases (U1163), 75015 PARIS

hassansaeiahan@gmail.com — hassansaei.github.io

#### **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

University of Tabriz, Iran

2013-2017

B.Sc. in Biology Overall GPA: 3.8

#### PROFESSIONAL EXPERIENCES

## **Imagine Institute of Genetic Diseases, Paris, France**

Jan 2025 - Present

Paris, France

Postdoctoral Researcher

· Research Focus: AAV targeted therapy development for podocytopathies in partnership with Sanofi, and White-Lab Genomics

Oct 2021 - Dec 2024

Ph.D. Candidate

Paris, France

· Research Focus: Improving genetic diagnosis in herediatry renal diseases and development of robust models for therapy development

#### ANALYTIC AND COMPUTATIONAL SKILLS

**Programing Languages** Python, R/Markdown, Bash, High Performance Computing, Git

**Workflow Managment** Docker, Singularity

**Big Data Analysis** RNA-seq, scRNA-seq, scATAC-seq, Proteomics

Sequence analysis GATK, deepVariant, CADD, SIFT, MutationTaster, IGV

Stem cell researchhiPSC Maintanance and DifferentiationDisease modelsOrganoids, Murine model Development

## 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 2<sup>nd</sup> place, National Biology Olympiad for undergraduate students, Ministry of Higher Education, Iran

<sup>\*\*</sup> All credentials are evaluated by WES

- 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. Therapeutic splice modulation of COL4A5 reinstates collagen IV assembly in an organoid model of X-linked Alport syndrome. *bioRxiv*. 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, Dorval G. Phenotypic Heterogeneity of ADTKD-MUC1 Diagnosed Using VNtyper, a Novel Genetic Technique *American Journal of Kidney Diseases*. 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 in silico 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)

# SELECTED SCIENTIFIC PRESENTATIONS

Section content...

# 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 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 Email: Talebi.s@iums.ac.ir