Report — Luiza Lorena Pires Ramos
Belgium to Sweden (October 2025)

Dear Committee Members,

I would like to express my sincere gratitude to the European Society of Human Genetics (ESHG) for granting me the opportunity to participate in the Observership Programme. I had the privilege of spending four weeks (6–31 October 2025) at the Department of Clinical Genetics and the Rare Diseases Research Group at Karolinska University Hospital and Karolinska Institutet, Stockholm, under the supervision of Professor Anna Lindstrand.

As a medical doctor specialized in Clinical Genetics and currently pursuing a PhD focused on rare unsolved neurogenetic disorders and long-read genome sequencing, I was particularly motivated to learn from an institution internationally recognized for its integration of advanced genomic technologies into clinical practice. The observership was structured into two complementary components that provided a comprehensive overview of both clinical and research activities.

During the first two weeks, based at Karolinska University Hospital, I shadowed the clinical genetics team in outpatient consultations and rounds, gaining a detailed understanding of clinical decision-making and patient management in a highly coordinated healthcare system. I also spent time with the clinical laboratory geneticists, where I learned about the diagnostic workflow used in routine practice and how it is embedded into Sweden's public healthcare framework. This included discussions on the ongoing efforts to implement long-read genome sequencing in the diagnostic setting—an area directly relevant to my own doctoral work.

The following two weeks were hosted by the Rare Diseases Research Group at Karolinska Institute. There, I had hands-on exposure to long-read genome sequencing data from the Undiagnosed Diseases Program. This experience allowed me to deepen my technical and analytical skills, particularly in the context of variant interpretation and the identification of unconventional genetic mechanisms. Several approaches and methodological insights acquired during this period can be directly applied to my current PhD project, strengthening my research pipeline and expanding potential avenues for investigation.

Beyond the technical and clinical learning, this observership offered a highly collaborative and welcoming environment. Interactions with clinicians, laboratory specialists, and researchers provided valuable perspectives on multidisciplinary practice, research translation, and healthcare organization. Observing how Clinical Genetics is practiced in a well-resourced setting—where advanced technologies, structured workflows, and coordinated patient pathways come together—was particularly inspiring, especially given my previous experience working in settings with more limited resources (Brazil).

I am sincerely grateful to Professor Anna Lindstrand for her generous supervision, as well as to the teams at Karolinska University Hospital and Karolinska Institute for their warm hospitality, thoughtful mentorship, and willingness to share their expertise. I also extend my appreciation to the European Society of Human Genetics for making this exceptional training opportunity possible.

This observership has been a highly enriching experience, contributing significantly to my academic development, clinical perspective, and research work. I enthusiastically recommend the program to other young geneticists seeking to expand their competencies in a stimulating and internationally collaborative environment.

Sincerely,
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