

ESHG Observership for Young Geneticists Programme 2025 – Final Report

Host Institution: Institute of Human Genetics, Jena University Hospital, Friedrich Schiller University, Jena, Germany
Supervisor: Prof. Dr. Thomas Liehr, PhD
Awardee: Sílvia Pires, MSc in Molecular and Cellular Biology; PhD candidate in Biomedical Sciences at ICBAS, University of Porto

Dear European Society of Human Genetics Education Committee,

As an awardee of the ESHG Observership for Young Geneticists Programme 2025, I had the privilege of completing a training placement at the Institute of Human Genetics, Jena University Hospital, under the supervision of Professor Thomas Liehr, a globally recognized expert in Molecular Cytogenetics. The institute is a leading reference center, offering a unique environment that integrates diagnostic services with genetics research.

During the observership, I gained comprehensive insight into the workflow of fluorescence *in situ* hybridization (FISH) methodology, from in-house probe preparation to its practical applications. I engaged with protocols involving chromosome microdissection, DOP-PCR amplification, probe labeling, tissue-FISH, and analysis of FISH results. This training not only deepened my understanding of FISH in routine diagnostic settings but also allowed me to incorporate the technique into my ongoing PhD research, which investigates the role of non-coding chromosomal heterochromatic regions in human health. I was able to apply advanced cytogenetic techniques, including the development of specific probe sets targeting human heterochromatin, to my doctoral project. The aim of this work is to bridge the gap between genetic variation and clinical outcomes, contributing both to scientific progress and to my academic development.

Beyond the technical aspects, the observership offered a rich multicultural and interdisciplinary experience. I collaborated with a highly skilled and welcoming team and engaged with colleagues from diverse international and academic backgrounds. These interactions broadened my scientific perspective, enhanced my understanding of current trends in genetics, and fostered a collaborative mindset.

I am sincerely grateful to Prof. Liehr and the entire team at the Institute of Human Genetics for their generosity, professionalism, and mentorship, and to the European Society of Human Genetics for making this exceptional opportunity possible. This observership has significantly enriched my academic training, personal growth, and professional development as a cytogeneticist.

Sincerely,

Sílvia Pires, MSc

Centro de Genética Médica Jacinto de Magalhães,
Unidade Local de Saúde de Santo António, Porto, Portugal
Instituto de Ciências Biomédicas Abel Salazar, Universidade do Porto, Portugal