Ana Rita Matos

The primary genetic cause and beyond: the life-time risk and phenotype modifiers in HDGC

Duration	4 weeks
Short Bio	I am a PhD student at the Institute for Research and Innovation in Health (i3S) in Porto, Portugal. My current research focusses on Hereditary Diffuse Gastric Cancer (HDGC), a deadly cancer syndrome associated with E-cadherin (CDH1) germline alterations. The research aims to understand why some CDH1 mutation carriers develop cancer early while others remain disease-free.
Home Institution	I3S – Institute for Research and Innovation in Health, Porto, Portugal
Host Institution	Centre Léon Bérard, Lyon, France
Project Description	This project focuses on HDGC syndrome and aims to understand the molecular mechanisms behind the varying risks within families carrying CDH1 mutations. The incomplete penetrance in HDGC families necessitates mutation-based risk assessments. The research is dedicated to predicting cancer development in CDH1 mutation carriers, enhancing decision-making for at-risk HDGC families. We seek to generate lifetime-risk estimations based on intra-familial or mutations-specific data. The project benefits from a 120-individual HDGC family's long-term study, with half carrying a CDH1-causing founder mutation. During the fellowship, I aimed to utilize a software package for cancer risk estimation developed by Youenn Drouet (Postdoc at Host Institute).
Personal Statement	Throughout the course of this fellowship, I discovered a newfound interest in bioinformatics and statistical analysis. Although I initially had a basic understanding of the tools and knowledge required to achieve our proposed tasks, the robust training I received equipped me with the necessary skills to delve into these complex fields. This experience not only broadened my expertise but also sparked a passion for the intersection of genetics, bioinformatics, and statistical analysis, laying the foundation for future pursuits in this dynamic and interdisciplinary field. I thoroughly enjoyed my experience during this fellowship. From day one, I felt consistently welcomed and embraced by Dr. Youenn Droet and his team. I was fortunate to have a host supervisor who was not only highly knowledgeable but also exceptionally supportive. Their availability and willingness to accommodate all my questions created a conducive environment for learning and collaboration, making my time in the fellowship immensely enjoyable and enriching.