Programa de Ayudas del GHEP-ISFG para Intercambios de Formación 2024-1

Title: Development of a microhaplotype panel for ancestry inference

Applicant: Pedro Manuel Fernandes Rodrigues

Institution of origin: Retsmedicinsk Institut, University of Copenhagen (UCPH)

Host intitution: Laboratório de Diagnóstico por DNA, Universidade do Estado do Rio de Janeiro (UERJ)

Exchange period: From the 27th of May to the 8th of August of 2024

Purpose of the trip and main results of the exchange:

As outlined in the grant application, we have been working towards the development of a microhaplotype panel for ancestry inference able to differentiate 5 metapopulations: Sub-Saharan Africa, Europe, South Asia, East Asia, and Native Americans. One of the main aims of this project is to sequence individuals from these regions. Therefore, it became particularly relevant to go to Laboratório de Diagnóstico por DNA (LDD), Rio de Janeiro, Brazil to prepare DNA libraries from Native American samples.

During the exchange period at LDD, amplification and library preparation was performed on 111 samples from Brazilian and Ecuadorian Native American populations for the Ion GeneStudio S5™ system (Thermo Fisher Scientific), which would later be sequenced at the institution of origin. The Brazilian samples were previously collected from Yanomami, Suruí, and Munduruku, all Amazonian groups, while the Ecuadorian samples represented Kichwa from both the Amazon and Andean regions. In addition, sequencing data generated at the institution of origin was analyzed and the necessary files for the analyses were prepared.

The data generated for these samples will be crucial in assessing how effectively the panel can differentiate Native Americans from other populations, or even differentiate among Native American populations from different geographic regions. Moreover, the analysis might uncover new variations within the microhaplotype region that are unique to these groups, enhancing the ancestry inference capacities of the panel. Besides the close work with my co-supervisor, this exchange allowed me to get to know other students and respective projects, as well as discuss ideas and share knowledge both in wet lab and data analysis. The findings obtained from this work will be submitted for publication as a scientific article.