



PhD project on **Genetic determinants of outstanding athletic performance** at IGBMC – Strasbourg - France

Research topic: World-class athletes display extraordinary physiological adaptations allowing them to perform at the limits of human endurance. These capabilities are driven not only by training and environment but also by genetic factors influencing muscle metabolism, oxygen utilization, and hypoxia tolerance. The 'Athlome' project aims to identify and characterize genetic variants associated with enhanced skeletal muscle performance under hypoxic conditions by analyzing the genomes of elite freedivers and ski mountaineers located at the extreme of the physiological performance distribution.

The PhD candidate will manage the ATHLOME project using advanced genomic and bioinformatic approaches. Whole-genome sequences from 44 elite athletes have already been generated, and the cohort will be expanded to approximately 80 world-class athletes, including international champions and record holders. Given the moderate cohort size, the project is specifically powered to detect rare variants with large functional effect, enriched in extreme performers compared to ancestry-matched reference populations (gnomAD and internal datasets). Extreme phenotype sampling substantially increases the probability of detecting rare variants of strong functional impact, conceptually analogous to monogenic disease genetics, compared to conventional population-based GWAS designs. In parallel, targeted gene-burden analyses will be conducted in genes and pathways previously implicated in oxygen transport, hypoxia signaling, and muscle performance, enabling detection of modest cumulative effects even within limited sample sizes.

Genotype–phenotype correlations will integrate detailed physiological parameters, including athletic and physiological performances and biochemical data. Functional validation of prioritized variants will be performed in vitro (splicing assays, protein stability analysis, enzymatic activity testing), providing mechanistic confirmation.

Understanding the genetic determinants of outstanding athletic performance in these unique extreme cohorts offers a natural human model of enhanced muscle and respiratory function. By focusing on rare, high-impact genetic determinants, ATHLOME aims to uncover protective molecular mechanisms potentially translatable to severe diseases characterized by muscle weakness and respiratory insufficiency, which are also actively investigated within our laboratory.

Candidate profile: you are a highly motivated and talented student with **expertise in Bioinformatics or Informatics** and a strong interest in sports science. English is the communication language in the team.

Location: The IGBMC institute is a main European center in Biomedical research equipped with 14 cutting-edge core facilities, and is a highly dynamic and international place hosting over 45 independent research groups working on diverse topics.

Strasbourg is a cosmopolitan city in a beautiful countryside, close to Germany and Switzerland and less than 2 hours from the center of Paris by train.

Application: send a pdf file with your CV and contact details for at least two referees to Jocelyn Laporte (jocelyn@igbmc.fr)
www.igbmc.fr/laporte