Postdoctoral Fellowship under the Marie S. Curie Actions Cofund project "Opening Sphere UAB-CEI to Postdoctoral Fellows (P-Sphere)" Gran Agreement 665919.

Department or Institution involved

Functional impact of human genome inversions in health and disease

Topic description

Despite the initial high expectations of genome variation studies, only a small proportion of the genetic risk of common and complex diseases has been identified. Inversions are one type of structural variant that affect a large part of the human genome and could have important consequences, but they have been poorly studied due to technical difficulties. As part of the INVFEST ERC Starting Grant, it has been possible to carry out the largest study of the frequency and distribution of inversions in humans, which provides an excellent opportunity to fill the existing gap in the knowledge of the functional effects of human inversions. In particular, the proposal aims to understand their role in disease and other traits relevant to health through several interrelated and interdisciplinary objectives: (1) Genotype a large fraction of human inversions in a high number of individuals well characterized at the phenotypic level or affected by several diseases using new methods currently being developed at the laboratory; (2) Associate the inversions with available functional data (gene-expression, epigenetic modifications) and other phenotypic information; and (3) Perform functional analysis of specific candidate inversions to dissect molecularly their potential effects. This will allow us to detect previously unknown associations of inversions with diseases and other important traits for health outcomes, which in turn could have a diagnostic value. Furthermore, it will provide information on the molecular pathways and interactions between risk factors involved in disease, which could help us improve prevention and treatment strategies within the personalized medicine framework.

Project supervisor & hosting group

Dr. Mario Cáceres, Comparative and Functional Genomics Group

The hosting group is one of the world leaders in the study of human inversions. Specifically, the group uses humans as a model and takes a multidisciplinary approach that combines new genomic methods and bioinformatic analysis of the great wealth of data available to generate results of interest to many diverse fields. To achieve this, it is formed by a young, dynamic, and interdisciplinary team of talented scientists who are experts in different areas. In addition, the group is focused both in basic science and translational research, which is exemplified by a significant scientific production with several high-impact publications and patents. As such, it has been awarded the prestigious ERC Starting Grant and Proof of Concept Grant for its work in the study of the inversions in the human genome.

Planned secondments

The research of this project will be carried out within the IN2DIS consortium, formed by different institutions of Estonia, UK, Netherlands, and Spain. As part of the research project, the candidate could carry out short research stages at the Estonian Genome Center, which harbours one of the largest human samples collections, to do the analysis of the data. It is also expected that additional international collaborations could be developed during the project depending on the results obtained.

Candidate's profile

We are seeking highly motivated and talented individuals with a PhD degree in areas related to genetics, genomics and molecular biology. Previous research experience in high-throughput genomic techniques, functional genomics, gene expression analysis, and molecular biology would be highly valued. Candidates should also have a good publication track record and be able to work well in a team environment.

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