# Position Offered: POSTDOCTORAL RESEARCHER

Project: Development of computational tools for the diagnosis of diseases caused by structural variants

Technological and scientific fields: Medical devices, computational biology, data analysis and integration

Location: Santander, Cantabria, Institute of Biomedicine and Biotechnology of Cantabria (IBBTEC), <a href="https://web.unican.es/ibbtec/en-us/">https://web.unican.es/ibbtec/en-us/</a>

Research Group/PI: Alvaro Rada Iglesias, Transcriptional regulation in development and congenital disease, https://web.unican.es/ibbtec/en-us/about-ibbtec/team/members/memberdetail?d=AlvaroRadaLAB

#### PROJECT SUMMARY

Structural variants (SVs) can cause disease through long-range mechanisms that affect the transcriptional regulation of genes. However, the prediction of long-range mechanisms is complicated and the etiological relevance of many SVs remains unknown. We recently developed POSTRE (Prediction Of STRuctural variant Effects), a computational tool that can predict the "long-range" pathomechanisms by which SVs cause certain types of congenital disorders. In this project we propose to analyze and integrate public single-cell genomic data (scRNA-seq and scATAC-seq) from human fetal tissues to generate regulatory maps in hundreds of human cell types. Once incorporated into POSTRE, these regulatory maps will allow us to (i) analyze the SVs involved in the majority of congenital defects and (ii) improve the sensitivity of POSTRE. Furthermore, this integration strategy will be also applied to single-cell genomic data obtained in adult tissues to extend the diagnostic use of POSTRE to common diseases (e.g. cancer).

#### PROFESSIONAL PROFILE

## Minimum requirements:

- Doctorate in Molecular Biology, Biomedicine or similar.
- Programming knowledge (R, Python or similar).

## Merits to be considered:

- Experience in the analysis and integration of differenty types of genomic data (e.g. RNAseq, ChIP-seq, ATAC-seq, Hi-C, etc.).
- Experience in prioritizing genetic variants involved in disease.

# WHAT IS OFFERED

We offer a postdoctoral contract in an internationally recognized group (e.g. ERC Consolidator Grant) and with a multidisciplinary and international composition. The project includes an extensive training program (240 ECTS) with diverse activities, including training internships in prestigious international laboratories (e.g. Prof. Malte Spielmann (University Medical Center Schleswig-Holstein, Germany).

### Contract conditions:

Indefinite contract for a Postdoctoral Researcher associated to the Momentum Project of 4 years' duration according to Spanish science law. Gross annual salary (41.000 € - 52.000 €).

Start of contract: before 31 December 2024

#### PRINCIPAL INVESTIGATOR CONTACT

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