Provisory Project Title: **Identification of genes responsible for Left Ventricular Noncompaction Cardiomyopathy: from bedside to the bench and back again**

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## Summary: (1000 characters)

Left Ventricular Noncompaction Cardiomyopathy (LVNC) is the third most prevalent human cardiac heart muscle disease characterized by extensive trabeculations and is presumably a failure of the normal myocardium compaction process during foetal development. Cardiac symptoms and manifestations of LVNC are heterogeneous, and might be aggravated with age. For most of the gene mutations associated with LVNC, a causal role in LVNC or a strong genotype-phenotype correlation remains unclear. In collaborations with an established network of Portuguese Cardiologist, we intend to identify novel genes involved in the onset and/or progression of LVNC that could be used as specific genetic markers to reliably diagnose LVNC and to refine the treatment of patients. We will reprogram cells from patients and control subjects (induced Pluripotent Stem Cells-iPSC) to establish cellular disease models, and use animal models to better understand the molecular mechanisms underlying LVNC pathology.

## Bibliographic references

(#indicates Joint first co-authors and * indicates corresponding author)

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