

**BioISI - Biosystems & Integrative Sciences Institute**

**VERÃO COM CIÊNCIA 2022 | BioISI Internship Projects**

**Project ID | VC22\_6**

**Project Title | Functional characterization of variants in the LDLR and APOB genes found in the Portuguese FH study**

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**Number of Fellowships available | 1 (one)**

**Abstract |** To perform functional study of variants of uncertain significance found in genes associated of FH. To study the variants in the LDLR, a directed mutagenesis will be carried out, the plasmid will be transfected into heterologous cells (involves cell culture) and then by flow cytometry the complete cycle of the LDLR will be evaluated. To study the variants in the APOB gene, a cell line that depends on cholesterol to grow will be used, which will be supplemented with LDL from patients and the LDLR cycle will also be studied by cytometry.