

Departamento Ciências da Vida

Human Genetics Unit

TEAM: Alexandra R. Fernandes (Assistant Professor), PhD
Students: Luis Raposo, Vanda Marques, Daniel Luis, Ana Silva, Fellows: Marina Pires (MSc), Ana Claudia Nunes (MSc), Joana Silva (MSc), Patricia Justiniano (MSc)



Susana Santos

- Degree: Biologia Microbiana e Genética, FCT-UL
- PhD: Biologia Molecular, FCT-UL
- Researcher at CQE-IST and DCV-UNL
- Auxiliar Professor FECCN - ULHT
- 15 papers
- 5 running projects

Objectives

- To understand the molecular and cellular mechanism underlying hypertrophic cardiomyopathy (HCM). In last instance to establish an accurate HCM genotype-phenotype correlation .
- To characterize the antiproliferative potential of organometallic complexes and to assess the biological targets by the use of tumor cell lines and animal models.
- To recognize the molecular mechanisms underlying canine mammary tumors.

Methodology

- Genotyping: Mass Spectrometry, High Resolution Melting (HRM) and Next Generation Sequencing (NGS)
- Transcriptional analysis and microRNA identification: Real Time PCR technology. RNA Seq; NGS.
- Cytotoxicity, cytoselectivity and cell death evaluation: MTS colorimetric assay, fluorescence microscopy and flow cytometry analysis.
- Affinity and interaction of DNA with the complex: UV titration and Electrophoretic Mobility Shift Assay (EMSA)
- Chromosomal aberrations identification: microscopy analysis.

Expected Results

- The genotyping strategy will allow the systematic detection of genomic variants in HCM. We hypothesized that sarcomere gene transcripts and specific microRNAs represent molecular markers of the typical HCM cardiac remodeling process. Also we intend to demonstrate the pathological effect of the identified mutations. This integrated strategy will provide insights into genotype/phenotype correlations.
- We demonstrate that some of the organometallic complexes synthesized at CQE-IST promote an in vitro cytostatic effect against colorectal and hepatocellular carcinoma and mammary gland adenocarcinoma cell lines. Proteomic analysis allow us to identify possible cellular targets that are being validated. These results are quite promising regarding the potential application of organometallic complexes in cancer treatment.

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