SCIENCESPRINGDAY

FACULDADE DE CIÊNCIAS E TECNOLOGIA
UNIVERSIDADE NOVA DE LISBOA

Department Departamento Ciências da Vida

Human Genetics Group

Research Unit / Team

Centro de Química Estrutural – Grupo V – Human Genetics

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

Partnerships

CEDOC Chronic Diseases FCM NOVA

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CI GMH Departamento de Genética

lpda LIGA PORTUGUESA DOS DIREITOS DO ANIMAL

Centro Veterinário Berna Tratar com afecto

Alexandra R Fernandes

•PhD in Biotechnology, IST, 2000

•Degree in Chemical Engineering (Biotechnology), IST, 1994

•35 publications in international journals with referrees

•8 FCT Projects (Team member); 1 European Project (Team member)

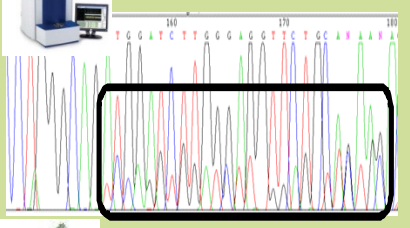
Objectives

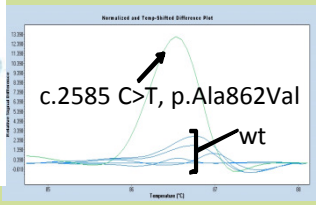
•To characterize the antiproliferative potential of organometallic compounds using tumor cell lines and animal models;

•To understand the molecular mechanisms underlying canine mammary tumors (through comparative proteomic analysis and RNA-seq)

•To establish genotype-phenotype correlations in hypertrophic cardiomyopathy (HCM) and to identify the miRNA profile associated with HCM progression.

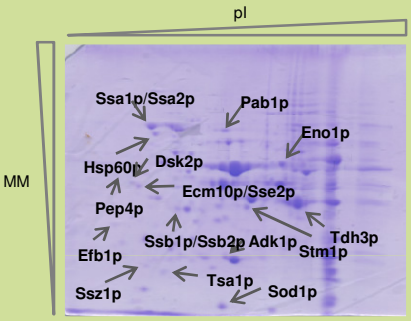
•Targeted therapy in cancer and cardiology





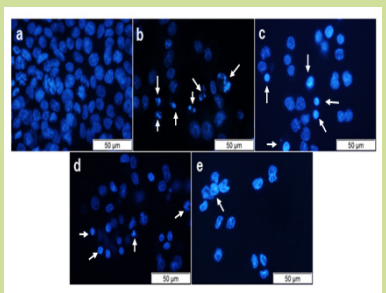
Methodology

- Genomic, transcriptomic and proteomic analysis (e.g. MassArray Maldi-TOF, real-time PCR, Next-Generation Sequencing, 2-D gel electrophoresis).
- Animal cell culture (primary and commercial human and canine cell lines);
- *In vitro* cell assays (cytotoxicity, cytoselectivity, cell cycle arrest, cell death evaluation) (e.g. MTS colorimetric assay, fluorescence microscopy and flow cytometry analysis); Identification of chromosomal aberrations (e.g karyotiping);
- *In vitro* DNA-organometallic compounds interaction studies (e.g. UV titration and Electrophoretic Mobility Shift Assay (EMSA), cleavage assays, religation assays);
- *In vivo* animal studies (for cancer therapy) (tumor size, fluorescence microscopy, immunohistochemistry).



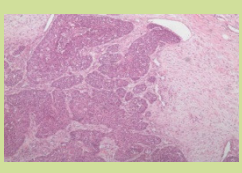
Expected Results

- Identification of new anti-tumor compounds with high specificity for tumor cells;
- Identification of biomarkers crucial for canine mammary tumors development and progression and amenable for targeted therapy;
- Identification of mutations associated with sudden cardiac death in hypertrophic cardiomyopathy patients and implementation of a clinical decision system based in the genetic and phenotypic data.



Recent publications:

Marta Gromicho et al., 2013. *Oncology Reports* 29(2):741-50.
Telma F.S. et al., 2012. *Dalton Trans* 41, 12888- 12897.
Santos, S., et al., 2012. *BMC Medical Genetics* 13:1.



Carcinoma type III with linfatic emboly

Funding: (Team member)
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FCT Fundação para a Ciência e a Tecnologia

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