

Procura-se candidato para concorrer a uma bolsa de Doutoramento na área da saúde

We're looking for a student to submit an application to the FCT call for PhD studentships

O concurso é o seguinte (FCT)/The FCT call for PhD studentships is at:

[https://www.fct.pt/noticias/index.phtml.pt?id=315&/2018/2/Concurso de Bolsas de Do
utoramento est%C3%A1 aberto at%C3%A9 28 de mar%C3%A7o](https://www.fct.pt/noticias/index.phtml.pt?id=315&/2018/2/Concurso%20de%20Bolsas%20de%20Doutoramento%20est%C3%A1%20aberto%20at%C3%A9%2028%20de%20mar%C3%A7o)

Prazo de candidatura/Application deadline: 28 março 2018/28th of March of 2018

O projecto é o seguinte/PhD project:

AntiSense Neurofibromatosis Therapy

The objective of this project is to investigate the development and potential application of the exon-skipping molecular approach in the context of Neurofibromatosis type 1, a rare disease. Neurofibromatosis type 1 is an autosomal dominant disorder that leads to a very diversified and apparently random set of clinical manifestations. It is presently considered to be the most frequent of rare diseases, with its prevalence ranging from one case in every 3,000 individuals to one case in every 6,000 individuals. Included among its most common manifestations are neurofibromas, low-grade gliomas, pigmentary abnormalities, learning and behavior disabilities, cardiovascular defects, and skeletal dysplasias. Although not always life threatening, affected people have its longevity shortened by approximately 10 years in average and can face many afflictions during their lifetime, such as deformities, and pain.

Although progress has been made in the last years, no completely satisfying therapy targeting the cause/origin of the disease has been obtained so far. One promising approach is based on antisense therapeutics and despite the success obtained with this approach in Duchenne muscular dystrophy no similar research has been carried out for the NF1 disease. Here, the main aim is to further the research in antisense therapy for NF1 and investigate the potential of exon-skipping for several types of NF1 mutations, some of them leading to stop codons in exons 36 or 37, which are some of the longest exons of the NF1 gene.

Responsáveis pela orientação/Supervisors: Dr. Sandra Alves from UID-Departamento de Genética Humana. Porto

Local de trabalho/Laboratório/Working place: UID-Departamento de Genética Humana. Porto

Applicants should have a master degree in Biology, Biochemistry or Biomedicine. Experience in Molecular Biology, RNA analysis, Reverse Transcription – Polymerase Chain Reaction are essential. Knowledge concerning western-blot is desirable. Applicant should send a cv, a recommendation letter with contact details and a cover letter detailing relevant experience to:

Dr Sandra Alves: sandra.alves@insa.min-saude.pt

and

Dr Ana Elisabete Pires: ana.alisabete.pires@gmail.com

Informal enquiries should be directed to Dr. Sandra Alves or Dr. Ana Elisabete Pires