

RESEARCH FELLOWSHIP (M/F)

Reference: PTDC/SAU-GMG/098305/2008

Title of the Project: Mapping and identification of new disease genes in autosomal dominant spinocerebellar ataxias

Internal Code: PR212003

A Fellowship is open for recruitment of a research fellow to collaborate in the Project referred above, financed by the Program “COMPETE - Programa Operacional Factores de Competitividade” in its FEDER component and by the Foundation for Science and Technology budget in its OE component

The fellowship is for one year, eventually renewable, starting on June 1st, 2010. The monthly amount of the fellowship is € 745,00.

Place of Work: Institute for Molecular and Cell Biology, Porto, Portugal

Work Program: See attached.

Description: The successful candidate will scan a 10K SNP array for linkage analysis by using statistical genetic software such as ALOHOMORA, SNPLINK and ALLEGRO.

Candidate profile: The candidate should possess a degree in Medicine, Pharmaceutical Sciences, Biomedicine, Biology, Biochemistry or related areas. We are looking for highly motivated candidates, having excellent academic marks/reports, with a deep interest in Mathematical and Human Molecular Genetics.

The applications should be received between May 12th and 26th, 2010.

Proposals must include a letter of motivation, CV, a letter of reference, and be sent to the e-mail candidaturas@ibmc.up.pt referring the internal code (PR212003).

The fellowship is regulated by current laws relating to the Statute of Science Research Fellows, namely Law 40/2004 of August 18, and the Regulation of Scientific Research Fellowships of the IBMC (www.ibmc.up.pt/fellowships.php).

Project: Mapping and identification of new disease genes in autosomal dominant spinocerebellar ataxias

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Summary:

Familial molecular studies have allowed a genetic classification of the autosomal dominant SCAs. We have been involved in molecular studies since 1991 (Silveira et al., 2002; Sequeiros et al., 2009; Sobrido et al., 2005). In Portugal, a systematic, population-based survey of hereditary ataxias was conducted for more than 12 years. Global prevalence of ataxias was 7.9:100,000 and prevalence of dominant spinocerebellar ataxias was described as 5.5, (Coutinho et al., 1994; Coutinho, personal communication, 2009). We have been active on mapping and cloning genes implicated in SCAs, together with mutation analysis which enabled the genetic characterization of approximately two hundred Portuguese families. However, there are still over one hundred families, and even more apparently sporadic cases, that lack a genetic diagnosis. In the Fundación Pública Galega de Medicina Xenómica (FPGMX; Santiago Compostela, Spain) over 300 unrelated ataxia patients have been received since 1997, for the genetic screening of SCA mutations. Similarly to what is observed in the Portuguese SCA population, about 50% of all cases sent for genetic diagnostics in Galicia remain undiagnosed. Therefore, we hypothesize that many Portuguese and Galician SCA families harbor unidentified mutations.

The successful candidate will perform research addressing the following:

- a) mapping new disease genes in Portuguese families by genome-wide linkage scan using a high density SNP array
- b) fine mapping within the linkage regions, using high-density SNP markers, for selection of candidate genes