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PROJECTE DE DOCTORAT INDUSTRIAL EXPEDIENT 2016 DI 013

DADES DE L'EMPRESA I DE L'ENTORN ACADÈMIC

Títol del projecte

Refining and improving a NGS gene panel for clinical diagnosis of hereditary hematologic diseases

Empresa

WHOLE GENIX S.L.

Responsable de l'empresa

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BREU DESCRIPCIÓ DEL PROJECTE DE RECERCA

Refining and improving a NGS gene panel for clinical diagnosis of hereditary hematologic diseases and discovery of new genes

The goal of this project is to improve an already established NGS-gene panel for rare inherited blood diseases in order to accelerate diagnostics. In addition, and due to our years of research we have identify several families with novel disease entities without a known causative gene. We propose to proceed with WES and WGS experiments in order to discover these causative genes. The candidate will be a person with a strong BIOINFORMATIC background and will focus on bioinformatics and computational work, as the experimental part is already covered in our laboratory.

Objectives:

- i) To refine and improve an already established Next generation sequencing (NGS) gene panel using emerging sequencing technology
- ii) Development of bioinformatics pipelines and optimization of the computational methods for faster analysis
- iii) Creation of an optimized platforms and databases.
- iv) Improve our LIMS system (laboratory database managing collection of samples system) with the program LIMS- Progeny.
- iv) Research studies by WES/WGS to identify new nosological entities in rare blood diseases



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Expected scientific and transfer results include:

- 1) Commercialization of a new and optimized NGS gene panel for rare hereditary hematologic diseases
- 2) Distribution of novel applications in the form of cloud-services in order to help medical doctors with the diagnosis and management of patients with these diseases.
- 3) Legal protected (patent) a novel database of variants in these diseases
- 4) The discovery of novel genes one new expectative for future development of new therapeutic strategies for these diseases

The employability plan encompasses the gain of knowledge in the clinical sector by the candidate during his/her PhD studies and surely the acquisition of skills that will be valued and demanded in the labour market.

This project will be developed at the Josep Carreras Leukaemia Research Institute, IJC (www.carrerasresearch.org) in the group of Dr Mayka Sanchez in close collaboration with the company Whole Genix and the Research Group in Bionformatics and Medical Statistics (2014SGR-596) of the University of Vic. The thesis will be codirected by Mayka Sanchez (IJC) and M.Luz Calle (Uvic-UCC).