Universitat Pompeu Fabra Barcelona

Master in Bioinformatics for Health Sciences

Master project 2021-2022

Personal Information

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Project

Computational genomics

Project Title:

Deciphering genetics of hereditary hemorrhagic telangiectasia

Keywords:

WES, variant calling, variant filtering and interpretation

Summary:

Hereditary hemorrhagic telangiectasia (HTT) is an autosomal dominant vascular dysplasia leading to epistaxis, telangiectasia and visceral arteriovenous malformations. Pathogenic variants in ENG and ACVRL1 are the main genetic cause responsible of the disease. Historically, genetic testing for HTT consisted of the analysis of ENG and ACVRL1. Nowadays whole exome sequencing (WES) has been introduced as diagnostic tool in patients with this disease. WES allowed the identification of several pathogenic genetic variants; nevertheless the proportion of unresolved exomes is much higher than expected. Particularly in HTT, in which the clinical phenotype is very specific, WES did not reveal, in the studied genes (ACVRL1, ENG, EPHB4, GDF2, RASA1 and SMAD4) , any pathogenic variant in 75% of patients. We assume the existence of other responsible genes o genetic mechanisms in HTT. The main objective of the project is to develop new algorithms for WES analysis in order to detect new candidate genes for HTT. This project will be jointly supervised with Dr. Irene Madrigal from the Molecular Genetics department at the Hospital Clínic de Barcelona, who is in charge for finding a genetic diagnosis for these patients.

Expected skills::

basic knowledge of human genetics, programming and analysis of next generation sequencing data

Possibility of funding::

No

Possible continuity with PhD: :

To be discussed