Laura Costa

Inherited Myopathies: Diagnostic value of clinical deep phenotyping, imaging and muscle biopsy in the era of Next Generation Sequencing

Short Bio	I am a child neurologist at Hospital Universitari Vall d'Hebron in Barcelona, with special interest in the clinical and research field of neuromuscular diseases. I am starting my PhD degree focused on the role of muscle imaging in the diagnosis and follow-up of neuromuscular patients and how it can give us a better understanding of the pathophysiology of the different neuromuscular diseases. I am also participating in clinical trials and research projects at Vall d'Hebron Research Institute in the Pediatric Neurology group with the aim of improving the diagnosis in neuromuscular diseases using omics technologies and machine learning.
Home Institution	Hospital Universitari Vall d'Hebron, Barcelona.
Host Institution	Ospedale Pediatrico Bambino Gesù, IRCCS, Roma
Project description	Inherited myopathies are genetically and clinically heterogeneous muscle disorders caused by mutations in a large number of genes. The incorporation of Next Generation Sequencing into clinical practice has completely changed the diagnostic procedure. In muscular disorders, although it is widely accepted that a precise clinical phenotyping is essential to guide and achieve the right diagnosis, questions have been raised about the value of muscle biopsy and wbMRI. The aim of this project is to study the success rate of molecular genetic diagnostic techniques applied to inherited myopathies in a reference center and how it is influenced by the precision and richness of phenotypic information. We will systematically collect data of patients with a suspected inherited myopathy in both institutions and by applying bioinformatic tools and simulations we will quantify the impact of each information on the diagnosis of a patient. Our final goal is to provide tools and clinical algorithms that enable a better definition of which patients and when in the diagnosis procedure will benefit from muscle biopsy and muscle imaging.
Personal statement	This is a great opportunity for me to improve my knowledge about the diagnostic procedure followed in a pediatric NMD reference center, gaining insight into the deep and precise phenotyping and NGS interpretation and validation. I also think it will enhance my skills in my daily clinical practice: recognition of specific phenotypes, identification of specific complementary tests adequacy (disease type and time point), and interpretation of NGS data.

	In addition, this collaboration will allow me to learn from a different way of clinical management and to enrich the ongoing research projects about related topics in my home institution and to strengthen the scientific relationship between both Institutions.