Elisa Vegezzi

Project title: *Genetic modifiers in hereditary ATTR amyloidosis*

Duration	6 months
Short Bio	I am a neurologist actively committed in the field of neurogenetics, with a particular interest in peripheral nerve disorders, especially in hereditary neuropathies. My Ph.D. program has been focused on the evaluation by quantitative muscle MRI of novel outcome measures in hereditary ATTR amyloidosis and on the clinical phenotyping and screening by whole-exome sequencing of a cohort of patients with suspected inherited neuropathies/ataxias/spastic paraparesis, trying to address the molecular diagnosis. More recently, I have started dedicating to the identification of genetic modifiers of age of onset and clinical phenotype of hereditary ATTR amyloidosis, stemming from a collaborative genome-wide association study (GWAS) project involving more than 50 Centres across the globe with a proven expertise on this rare condition, which has been the topic of my ERN Research Mobility Fellowship
Home Institution	Mondino Foundation IRCCS (Pavia)
Host institution	UCL Queen Square Institute of Neurology (London)
Project description	The project is aimed at identifying loci harboring genetic variations which modify age of onset, penetrance, phenotype, clinical severity, progression, and response to anti-amyloidogenic treatments in hereditary ATTR amyloidosis through a GWAS. Also, it will characterize by long-read sequencing (*PacBio* and *Oxford Nanopore*) the *TTR*-containing region across different populations and ethnicities to phase nearby variants (*cis* or *trans*) associated with the *TTR* mutation itself which define different intragenic haplotypes
Personal statement	This exchange gave me the opportunity to deepen my knowledge of GWA studies and data analysis, and to gather specific expertise on long-read sequencing technique (*PacBio* and *Oxford Nanopore*), which, beyond the specific case of hereditary ATTR amyloidosis here reported, could be also applied to other inherited neurological disorders to better explain their genetic bases, or even to acquired conditions. Also, since the host Institution (UCL Queen Square Institute of Neurology) is member of both EURO-RND and EURO-NMD and has proven expertise on GWA studies, the development of the project on hereditary ATTR amyloidosis in this context, has provided so far

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	seminal tasks/methodology to run similar research projects also for different rare neurological disorders

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