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## Stephanie Efthymiou

*Improving our understanding of the global genetic architecture and phenotype of FSHD*

Duration	1 month
Short Bio	I am a Post-doctoral Research Fellow and Senior Technician for the International Centre for Genomic Medicine in Neuromuscular Diseases (ICGNMD). My current research focus is studying genetic basis of neurodmuscular disorders seen in patients from different ethnic groups. Next-generation sequencing technology together with deep phenotyping and functional studies are all important aspects of my research.
Home Institution	UCL Queen Square Institute of Neurology
Host Institution	Leiden University Medical Center
Project Description	FSHD is the third most common muscular dystrophy causing progressive muscle weakness. Most of current FSHD knowledge is based on European and Asian populations and data are lacking from other ethnic background which is detrimental to the future development of diagnostics and interventions. The aim of the ICGNMD project is to engage with low-to-middle income partners who have no in-country diagnostic or research capability to genetically characterise FSHD. The aim of the mobility fellowship was to learn the FSHD testing protocol from Prof. van der Maarel's research group. Subsequently, the aim was to use this collaboration to provide more training and build on the engagement with ICGNMD partners to improve our understanding of the global genetic architecture of FSHD patients.
Personal Statement	The EJPRD fellowship has established a valuable exchange between ICGNMD and LUMC which has offered me valuable transferable skills and new insights on translating medical discovery in FSHD. Meeting new researchers and exchanging ideas has cultivated my scientific thinking and collaborative mindset but also advanced my career as a medical researcher. An FSHD testing lab has been set up at UCL which uses optical genome mapping to complement the Southern-blot based diagnosis carried out at Leiden, therefore building on a long-term collaboration between Prof. Van der Maarel's group and relevant ICGNMD partners.

