Daniel López Domínguez

Project title: Neurofilament light chain as a biomarker for Spinocerebellar ataxias. Study of its correlations with motor, cognitive, and radiological parameters.

Duration	3 months.
Short Bio	I completed my degree in Medicine at the University of Santiago de Compostela, and later specialized in Neurology (3 years ago) at the Josep Trueta Hospital, in Girona, Spain. During the residency I had a special interest in Movement Disorders, Ataxias and Neurogenetics, and after its completion I have been part of the Movement Disorders Unit and Ataxias Unit, with participation in own and external studies, especially in relation to Parkinson's disease, ataxias, etc.
Home Institution	Hospital Josep Trueta, Girona – Hospital Santa Caterina, Salt. Spain
Host institution	Hôpital de la Pitié-Salpêtrière, Paris. France
Project description	SpinoCerebellar Ataxias (SCAs) are a heterogeneous group of hereditary pathologies. The most frequent forms share the same pathogenic mechanism: the expansion of CAG triplets encoding elongated polyglutamine tracts. The clinical picture includes cerebellar syndrome, movement disorders, etc. Neurofilaments light chain (NfL) have been described as a promising biomarker on different neurodegenerative diseases, showing promising results in SCA as well. On the other hand, the Cerebellar Cognitive Affective Cerebellar Syndrome (CCAS) has recently been described as a specific cognitive affection secondary to cerebellar injury. Although it has been described in various forms of SCA, its study has not yet been carried out in large cohorts. Therefore, the objective of the project is to study the presence of CCAS in the most common forms of SCA, as well as its correlation with motor symptoms, NfL and neuroimaging (MRI) markers.
Personal statement	The EJPRD fellowship has been a great experience for my career as a neurologist, and the fact that I was able to observe the largest

	research projects (at European and global level) and the minds working behind them on a reference center was a great learning opportunity. It has been very enriching to have been able to develop a project with the experienced and dynamic team directed by Dr Durr. Their knowledge and way of working have clearly improved my approach in the Neurogenetics consultation and will influence my academic work in the coming years, as a first step towards the development of my PhD. Above all, I would highlight the importance of their multidisciplinary approach, enriching both for the rare diseases care and research, as well as the importance of the team, to solve both the simple and complex cases.