Ana Daniela de Oliveira e Silva

Project title: *Clinical and molecular characterisation of BAF complex-related genetic syndromes*

Duration	*6 months*
Short Bio	*Neurodevelopmental disorders affect 1-3% of the world population causing a tremendous socio-economic impact. Genetic factors play a key role in the etiology of these neurodevelopmental disorders. Nonetheless, these pathologies are genetically characterized by a significant allelic and locus heterogeneity. The SWI/SNF complex [ATP-dependent BRG1/BRM associated factor (BAF)] is a chromatin remodelling multiprotein complex that plays a crucial role in gene expression regulation and neural development. It is essential in epigenetic regulation through nucleosome remodelling. Mutations in genes that encode BAF-complex subunits cause a spectrum of mendelian neurodevelopmental disorders with some phenotypic overlap, commonly known as BAFopathies.*
Home Institution	*Coimbra Hospital and University Centre*
Host institution	*Guy's and St Thomas' NHS Foundation Trust*
Project description	*The project "Clinical and molecular characterisation of BAF complex-related genetic syndromes" aims to perform clinical and multi-omics characterisation of BAF complex-related genetic syndromes in order to provide a better understanding of the molecular basis, pathophysiology, and phenotypic variability of these disorders in the Portuguese population. There will be a particular focus on growth, psychomotor development, epilepsy and other neurologic complications.*
Personal statement	*This fellowship allowed me to deepen my knowledge on this topic which has been crucial for the development of a comprehensive characterisation of a Portuguese cohort of patients with BAFopathies regarding their phenotype and genotype and whose results we have been sharing in different scientific meetings. I had the opportunity to attend specialized clinics at the Department of Clinical Genetics (Guy's and St Thomas' NHS Foundation Trust) and participate in clinical preparation and notes review of cases with different diagnosis including chromatinopathies (in particular BAFopathies). This experience was extremely important to develop my knowledge about this group of pathologies and understand how these patients are clinically*

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	managed in this department. I also had the opportunity to develop my skills in interpretation of laboratorial results and in basic science as I worked in close contact with a research group focused on neurodevelopmental disorders – RDND lab (King's College London). It was a remarkable opportunity to learn and develop skills from clinical and laboratorial experts in this area.

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