Understanding the Genetic Basis of Dementia with Lewy Bodies

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Name of Fellow

Dr Jose Bras

Institution
Funder

Alzheimer's Society

Contact information of fellow
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United Kingdom

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The project/programme is most relevant to:

Alzheimer's disease & other dementias

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Research Abstract

Background: Dementia with Lewy Bodies (DLB) is a complex brain disorder that has been severely underserved and whose etiology remains unclear. A growing body of evidence supports the notion that Alzheimer’s disease (AD), DLB, and Parkinson’s disease (PD) are members of the same disease continuum.

I have collected, through collaborations, the world’s largest cohort of DLB cases, currently over 1,000 samples, of which 764 are neuropathologically diagnosed.

Hypothesis: We hypothesize that there is a genetic component to DLB including both rare causal and low-frequency risk variants.

Specific Aims: To perform exome-sequencing in a total of 764 neuropathologically confirmed DLB cases to identify rare causal protein-coding mutations; to fine-map previously identified associations to pinpoint risk-conferring variants; to make these data available to the scientific community.

Study Design: I will perform exome-sequencing of neuropathologically confirmed DLB cases and, simultaneously, sequence the genomic regions underlying association signals. These data will be used to impute genotypes in clinically diagnosed samples. Using both publicly available and in-house controls, I will identify disease-specific and risk-conferring variants.

Relevance: Understanding the genetics of DLB has the potential to inform regarding pathobiological events and ultimately aid in recognizing points of therapeutic intervention and in the identification of at-risk individuals. Although not a primary goal, this study also has the potential to uncover genetic convergent points between AD, DLB and PD.

Expected Outcomes: I will pinpoint causative mutations for DLB, and identify variants that modulate the risk of an individual to develop this disorder.

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