

Capacity building in sub-Saharan Africa to conduct cutting-edge genetics research in Parkinsons disease.

<https://neurodegenerationresearch.eu/survey/capacity-building-in-sub-saharan-africa-to-conduct-cutting-edge-genetics-research-in-parkinsons-disease/>

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USA

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Capacity building in sub-Saharan Africa to conduct cutting-edge genetics research in Parkinsons disease.

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2

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Research Abstract

PROJECT SUMMARY/ ABSTRACT Parkinson's disease (PD) is estimated to affect over 7 million people worldwide. There is no cure and current treatment only alleviates the symptoms and do not stop neuronal loss. Due to the array and severity of motor and non-motor symptoms

experienced by PD patients this has a significant effect on their quality of life as well as that of their caregivers. Further studies are urgently needed on the risk factors that place certain individuals at higher risk for PD development. It has been shown that PD has a significant genetic component and a number of genes have been discovered however these studies were predominantly done on Caucasian and Asian populations. To date, there have been fewer than 10 published genetic studies on PD in Black populations. It is plausible that these patients harbor pathogenic mutations in novel PD-causing genes. The discovery of genes such as parkin and PINK1 have shed light on possible disease mechanisms underlying PD including mitochondrial dysfunction, mitophagy and oxidative stress, and hence identification of other novel PD genes has the potential to reveal important insight into the pathobiology of the disorder. It is therefore imperative that more studies are done on Black PD patients. In the proposed study we shall examine the hypothesis that Black PD patients harbor mutations in novel PD-causing genes due to their unique ancestry. This R21 project aims to build collaborative research projects between researchers in Nigeria, South Africa and the USA to study the genetic causes of PD of Black patients from these three countries. We aim to recruit a minimum of 100 Black PD patients in Nigeria and South Africa for genetic studies, screen for pathogenic mutations in the known PD-causing genes as well as a panel of 751 neurologically-associated genes. Moreover, we will set up a pilot study to screen for mutations in mitochondrial DNA. Finally, we will compare and contrast the clinical and genetic findings across the three populations and to that of other published studies. Over the two years of the project, research capacity on PD in Nigeria will be developed through setting up the infrastructure for patient recruitment, follow-up and clinical assessments. Capacity in South Africa will be developed through the focussed recruitment of Black PD patients, and the setting up of high-throughput targeted re-sequencing approaches. Capacity in the USA will be developed through access to large numbers of Black patient numbers and the setting up of the mitochondrial DNA sequencing approach. It is anticipated that the preliminary data produced through this research collaboration will form the basis for a future larger- scale R01 grant application. The overall impact of this study is that by identifying novel PD genes this will shed light on disease mechanisms underlying neurodegeneration and ultimately this will inform the design of more effective therapies for not only PD but also other forms of neurodegeneration.

Further information available at:

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