Identification of causal genetic mutations for the early diagnosis of Parkinson's Disease

Parkinson’s Disease (PD) is a debilitating brain disorder that leads to difficulties in movement, notably tremors at rest. The protein a-synuclein has been identified as a possible site of mutation and its role in PD has garnered significant interest. Whole-transcriptome brain gene analyses have long shown the potential to provide insight into the causal molecular pathways associated with brain disorders such as PD. A common hurdle to this approach, however, is the likelihood that analyses will produce ‘culprits’ that are often only secondary to disease pathology and not indicative of the underlying cause of disease. This technology focuses on the a-synuclein protein and establishes a differential analysis that highlights the absolute amounts of a-synuclein within the context of holistic measures. This transcriptome wiring analysis provides a potentially causative relationship between a-synuclein and Parkinson’s Disease pathogenesis.

Provides a highly sensitive measure for the investigation of a-synuclein as a potentially causal player in Parkinson’s Disease pathogenesis.

A unique isoform of a-synuclein may shed light on the pathogenesis of PD. This technology quantifies the level of this unique isoform with respect to holistic levels of a-synuclein, a measure that has correlated highly with PD. In this way, it may be possible to elucidate a biomarker in the pathogenesis of disease and provide an avenue for therapeutic intervention.

Published reports using this technology suggest that a-synuclein and indicators of PD are co-localized and highly correlated, confirming its potential use as a biomarker of disease.

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Applications:

- Identifies genetic biomarkers that could aid in the diagnosis of Parkinson's Disease.
• Potential application as a pathological identifier in other diseases that result from transcription errors or neural degeneration.

**Advantages:**

• Identifies causal factors of Parkinson’s Disease and provides a potential biomarker for the diagnosis of disease as well as therapeutic approaches.
• Employs a methodology that can serve as a platform for the study of multiple other diseases, including other neurodegenerative disorders.

**Patent information:**

Patent Pending ([WO/2013/086041](WO/2013/086041))

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**Related Publications:**


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