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Examining Discordant Twin Pair for Progressive Supranuclear Palsy: Genetics, MRI and Risk Factors

Dora Nowak*

Department of Genetics, Semmelweis University, 1085 Budapest, Hungary

Introduction

Progressive Supranuclear Palsy (PSP) is a rare neurodegenerative disorder characterized by a spectrum of clinical features, including balance and mobility impairments, cognitive decline and oculomotor dysfunction. Although there is a growing understanding of the disease, its etiology and pathophysiology remain elusive [1]. Twin studies offer a unique opportunity to explore the role of genetics, environmental factors and biomarkers in the development of PSP. In this investigation, we delve into the case of a discordant twin pair, where one twin developed PSP while the other remained unaffected. This rare scenario provides a valuable lens through which we can examine genetic and environmental risk factors, neuroimaging changes and blood-based markers associated with PSP. The objective is to unravel the complex interplay between genetic susceptibility, environmental influences and biomarkers in the context of PSP, aiming to shed light on disease mechanisms and potential diagnostic and therapeutic avenues [2].

Description

This study focuses on a remarkable case involving identical twins discordant for PSP. The affected twin was diagnosed with PSP based on clinical presentation, while the unaffected co-twin serves as a unique control in the investigation [3]. Genetic analysis aims to explore the presence of known risk variants associated with PSP and identify potential genetic modifiers contributing to the discordance. Additionally, neuroimaging with Magnetic Resonance Imaging (MRI) is utilized to discern structural and functional brain changes associated with the disease and their potential role in the clinical divergence of the twins [4]. Blood-based biomarkers, encompassing neuroinflammatory markers, tau protein levels and oxidative stress indicators, are also scrutinized to understand their relevance to disease onset and progression. Lastly, the study delves into environmental and lifestyle factors, seeking to identify potential triggers or protective elements contributing to the differences in disease manifestation between the twins [5].

Conclusion

The examination of this discordant twin pair in the context of progressive supranuclear palsy sheds light on the multifaceted nature of this enigmatic neurodegenerative disease. While genetic factors may set the stage for susceptibility, the disease's progression appears to be influenced by a complex interplay of genetic modifiers, environmental factors and potential protective mechanisms. Neuroimaging and biomarker analysis offer valuable insights into the brain changes and molecular signatures associated with PSP. By dissecting the intricacies of this discordant twin case, we aim to advance our understanding of PSP, potentially paving the way for improved diagnostic strategies, targeted therapies and a more comprehensive view of neurodegenerative disorders as a whole. This investigation underscores the significance of multifaceted research approaches in unravelling the mysteries of complex neurological conditions like PSP.

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Conflict of Interest

There are no conflicts of interest by author.

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***Address for Correspondence:** Dora Nowak, Department of Genetics, Semmelweis University, 1085 Budapest, Hungary, E-mail: dnowak@hotmail.com*

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