

Good example of parkinsons disease research paper

[Science](#), [Genetics](#)



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Introduction

Parkinson's disease (PD) refers to a chronic progressive neurodegenerative disorder. It is the second most predominant neurodegenerative condition after Alzheimer's disease (Thomas & Beal, 2007). Advanced age increases the risk of disease. It is diagnosed in 1% of individuals with 65 years of age and 5% of the individuals with over 85 years (Weintraub, Comella, & Horn, 2008). However, up to 3% of the cases have been found in people below 50 years. Other risk factors include the male gender and individuals of European ancestry. PD is a chronic disorder that progresses slowly; it may take 15 years from the time the disorder is diagnosed to death. It is mainly characterized by motor manifestations and loss of midbrain dopaminergic cells. PD also causes a range of non-motor symptoms and pathology in a number of regions of the nervous system. The onset of symptoms may

appear before PD can be clinically recognized. Research studies have identified genes such as α -synuclein and molecular pathways such as mitochondria impairment that are responsible for PD. However, other studies postulated that environmental factors could increase the risks of disease development. Current therapies provide symptomatic relief but fail to halt disease progression. Poor understanding of disease mechanisms has greatly hindered the development of neuroprotective therapies.

Etiology

The principal cause of PD is destruction of the dopamine-producing tissues in the substantia nigra as a due to the interaction between genetics and environmental toxins (Gwinn, 2013). Some pesticides have been shown to double the risk of developing the disease. The herbicide paraquat which is a mitochondrial toxin has been linked to dopaminergic cell loss in experiment animal models. Heavy metals such as manganese are thought to increase the incidences of PD. However, these hypotheses lack epidemiological support (Shulman, De Jager, & Feany, 2011). Research studies based on diets and habits have indicated that cigarette smoking and coffee reduce an individual's susceptibility to PD (Gwinn, 2013).

Pathology

The principal neuropathological trait is the loss of substantia nigra dopaminergic cells and the subsequent development of protein-rich inclusions known as lewis bodies (Shulman, De Jager, & Feany, 2011). Lewis bodies (LB) are spherical eosinophilic proteins that stain for α -synuclein protein. The brains of PD patients show accumulations of α -synuclein within

the neuronal processes with more diffuse staining patterns. Since dopaminergic cells contain melanin, loss of these cells causes depigmentation of the midbrain..

Pathogenesis

PD has been linked with gene mutations found in chromosome 4 (Weintraub, Comella, & Horn, 2008). Several genes have been associated with PD; SNCA gene, LRRK2, MAPT and GBA (Thomas & Beal, 2007). The SNCA that maps chromosome 4q21 forms a major pathway that is involved in neurodegeneration in PD (Shulman, De Jager, & Feany, 2011). PD patients exhibit missense mutations in the SNCA gene. These are A30P, A53T and E46K (Thomas & Beal, 2007). These mutations lead to increased gene expression through locus multiplication or polymorphism of promoter sequences triggering the toxic pathway. This results in disease onset in the 4th or 5th decade of life and development of rigidity and bradykinin. Autopsy reveals α -synuclein pathology in the neocortex, brainstem and limbic areas and loss of dopamine-producing cells in the SN. The other PD-susceptible genes MAPT, LRRK2 and GBA, promote this cascade. GBA causes degradation of lysosome that has been linked to PD. However, extensive research is necessary in order to understand the intricate mechanism (Shulman, De Jager, & Feany, 2011). Susceptibility is further aggravated by aging and environmental factors. Some environmental toxins target and damage the mitochondria causing production and accumulation of reactive oxygen species.

Clinical Presentation

The principal clinical features are resting tremor, rigidity, bradykinesia, gait impairment and postural instability. Besides the motor dysfunction and dopaminergic cell loss, recent research has identified non-motor symptoms and pathological features in the nervous system (Shulman, De Jager, & Feany, 2011). These features include disordered sleep, impaired olfaction and constipation, and they precede the motor symptoms by as much as 20 years. Advanced PD is typified by frequent motor freezing and falls, dyskinesias, pain and sensory complaints, neuropsychiatric symptoms and autonomic dysfunctions.

Treatment

Currently, there are no therapies that can alter the neurodegenerative process appreciably, and physicians rely on supportive therapy. Dopamine replacement therapy is used to manage the motor symptoms. Non-motor features are not responsive to dopamine replacement, and they play a fundamental role in overall disability.

Conclusion

PD is an intricate condition with multiple causes whose mechanism in causing PD is poorly understood. This has greatly affected discovery of therapeutic interventions that would halt disease progression. Current research has focused predominantly on the vulnerability of dopaminergic cells in the substantia nigra creating a knowledge gap as to how the disorder spreads in other brain systems. Further research is required to elucidate the involved pathways and to identify possible targets for therapy.

Reference List

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