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January 19, 2021

We Can Perform Early Screening for Neurodegerative Disorders? Does That Mean We Should?

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In the November 2020 issue of Movement Disorders, Dommershuijsen and his fellow researchers in the Netherlands wrote an excellent editorial on the ethical considerations of screening for rapid eye movement sleep behavior disorder (RBD) in the healthy general population. I wanted to use this blog to discuss why this question is such an important one to consider as discoveries in the alpha-synucleinopathies continue to advance.

We now know that there are five distinct disorders caused by inappropriate accumulation of alpha-synuclein within nerves of the central and peripheral nervous system: Parkinson's disease, dementia with Lewy bodies, multiple system atrophy, pure autonomic failure, and RBD. It is also clear that some of these disorders may be prodromal illnesses for other diseases within the family. For example, work by physician-researcher Horacio Kaufmann has shown that almost 90% of people with pure autonomic failure will go on to develop other neurodegenerative diseases within 15 years. It has also been shown by Posthuma and others that up to 90% of patients with RBD will go on to develop other synucleinopathies, most often Parkinson's disease, over a 10- to 20-year period. These observations, although incredibly elucidating for clinicians, can be frustrating and scary for patients.

At a research level, the ability to identify patients who are 10-20 years from developing the clinical features of a progressive neurodegenerative disease opens up all types of opportunities. Instead of allowing these synucleinopathies to develop for 20 years by building up synuclein within the nerves and then hoping to identify a therapeutic agent that can reverse the process, the ability to identify misfolded synuclein when patients have only minimal symptoms introduces the possibility of neuroprotection—a mechanism through which we can prevent diseases before they occur in people who we know are at high risk.