Roy Alcalay MD, MS

Assistant Professor of Neurology
Division of Movement Disorders

Parkinson's disease is very heterogeneous - some patients progress slowly while others decline rapidly; some exhibit cognitive deficits and others suffer from postural difficulties; and the age of onset can vary considerably. Roy Alcalay suspected that these phenotypic differences could be traced back to different genetic mutations. In collaboration with Dr. Karen Marder and Dr. Lorraine Clark, he has performed analyses comparing mutation carriers to non-carriers indicating that this is in fact the case.

Now, he wants to explore the low penetrance of a couple of these mutations and find biological markers to aid in distinguishing those carriers who will go on to develop Parkinson's disease from those who will not. He is in the process of screening carriers for non-motor symptoms that are known to predate the onset of full blown Parkinson's disease, like REM sleep behavior disorder, a decreased sense of smell, cognitive changes, lowered blood pressure, and constipation. Once a pharmacological intervention is found this cohort will be ready and willing to test it, since by the time Parkinson's sets in it is often too late to intervene. It is still not known exactly how these mutations cause Parkinson's disease, and Dr. Alcalay is collaborating with certain basic labs (focusing on the lysosome) to explore the mechanism of the risk for Parkinson's in these mutation carriers.

Research focus: identifying genetic risk factors and biomarkers for the varying phenotypes of Parkinson's disease
Resources/ expertise: a repository of DNA, plasma and spinal fluid samples from people with mutations in the GBA gene, both those who have developed Parkinson's disease and those who have not
Resources wanted: biological markers for Parkinson's disease, both for more accurate diagnosis and to monitor disease progression, since now the diagnosis is made on a purely clinical basis.

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