

4. Annemarie Opprecht Parkinson Award 2008

Parkinson's disease: where are we up to in understanding the causes of the disease and how does this work fit in?

Statement for lay people

Parkinson's disease (PD) is a chronic progressive neurodegenerative disease without any known cure. The economic, social and public health burden of PD will increase as the World population ages.

Scientific investigation of forms of Parkinson's disease that run in families have led to the successful identification of five genes linked to this debilitating disease; however, even collectively these mutations account for only a few percent of cases. In an attempt to begin to find common genetic variation that may alter lifetime risk for Parkinson's disease in a large proportion of people with the disease we used a new technology that permits analysis of hundreds of thousands of points within the human genome in parallel. The idea behind this work was primarily to identify genetic risk variants that are common and strong influence on risk for disease, but also to produce data that could be used and added to by other scientists in the field of Parkinson's disease research and other related disorders.

The results of this work suggest that there is no single common genetic risk factor that has a strong effect on disease but does not preclude the possibility that the disease results from many genetic variants interacting to bring about the disease. The data generated by this work were the first to be placed into the public domain and to date have been accessed by several hundred research groups.

Dr. Singleton and Hardy's groups are currently expanding upon this work to find risk variants in Parkinson's disease, with the ultimate aim of defining risk variants that may identify those at risk for disease in addition to improving understanding of the disease process.

In addition Drs. Hardy and Singleton previously found that overproduction of the synuclein protein caused one genetic form of the disease and Dr. Singleton was a co-discoverer of the dardarin gene, a common cause of Parkinson's disease, accounting for up to 10% of cases in some populations.

Reading List:

Fung H-C, Scholz S, Matarin M, Simon-Sanchez J, Hernandez D, Britton A, Gibbs JR, Langefeld C, Stiegert ML, Schymick J, Okun M, Mandel RJ, Fernandez HH, Foote KD, Rodriguez RL, Peckham E, Wavrant De Vrieze F, Gwinn-Hardy K, Hardy JA, Singleton AB (2006) Genome Wide Genotyping in Parkinson's Disease and Neurologically Normal Controls; First Stage Analysis and Public Release of Data. Lancet Neurology. 5: 911-16