Lay Summary

BRAIN UK Ref: 19/007

Pathological and genetic study of an unusual case of alpha-synucleinopathy

Prof Janice Holton, University College London

In a group of diseases known as α-synucleinopathies an abnormally sticky protein called α-synuclein forms clumps inside brain cells causing nerve cells to die. The most common disease in this group is Parkinson's disease (PD) but there is also a rarer form called multiple system atrophy (MSA). PD and MSA do not usually occur in families but there are rare instances of familial PD caused by alteration (mutation) in the gene that codes for α-synuclein protein. One clue that there may be a mutation in this gene is the finding of an unusual pattern of α-synuclein inclusions when the brain is examined. We have observed an unusual pattern of these changes in a single case and would like to investigate this further to determine whether it is due to a gene mutation. Study of rare cases has often enabled us to have a better understanding of common diseases. We hope that by performing a detailed study of pathology and genetics in this single case we may gain insight into the mechanisms causing PD and MSA. This would be very important for this group of people who have progressive disease with no currently available treatments that can alter the disease course.