Lower motor neuron pathology in Down's Syndrome
Kalmar, B., Greensmith, L., Fisher*, E.
*Chief Investigator.

UCL Institute of Neurology, Queen Square, London, WC1N 3BG, United Kingdom (Email: e.fisher@prion.ucl.ac.uk Tel: +44 203 448 4439)

Research question and background
Down’s syndrome is a complex multisystem disease. It has been long known that some parts of the motor system affecting balance and coordination, such as the substantia nigra and the cerebellum are affected in the disease and neurons are lost in people with Down’s syndrome (DS). Although it has been long known that people with DS have reduced muscle force, there is no evidence of pathology occurring in the spinal cord, affecting lower motor neurons. Thus, in this study we asked whether we can detect spinal cord motor neuron loss in people with Down’s syndrome.

Methods and tissues used
We requested cervical spinal cord samples from people with i) Down’s Syndrome, ii) Alzheimer's Disease; iii) Amyotrophic Lateral Sclerosis and iv) non- neurological controls. Each specimen block was processed and cut to collect 20 sections, which are each 60 µm apart. Sections were then stained for Nissl in order to visualize neuronal cells. The number of large motor neurons was established in the ventral horn of all 20 sections in each sample.

Results and conclusion
A comprehensive quantification of the number of motor neurons in all samples we received has been completed. Our results show that there may be a loss of spinal cord motor neurons in people with Down’s syndrome. We are currently analysing our mouse model data to complement the human study, and we expect this analysis to continue into 2015 and possibly beyond.