Research Project:

Balance and gait abnormalities in adult patients with mitochondrial disease and spinocerebellar ataxia type 6

Principal researcher: Dr Yi Shiau Ng, Wellcome Centre for Mitochondrial Research, Newcastle University (UK)

Scientific Summary

Two-thirds of adult patients with mitochondrial disease demonstrate features of cerebellar dysfunction such as unsteady gait, dysarthria and uncoordinated limb movement. Patients with ataxia are at high risk of recurrent falls and injuries leading to frequent medical visits.

Laboratory gait and balance assessments are emerging as promising tools to characterise and track spatial-temporal gait changes in cerebellar ataxia. More recently, instrumented gait analysis has been able to identify gait abnormalities in adult patients with mitochondrial disease compared to healthy controls. However, patients with mitochondrial disease often have co-existing neurological features such as vestibular dysfunction, complex ophthalmoplegia, fatigue, myopathy and neuropathy, which often contribute to balance difficulties and gait impairment, in addition to cerebellar dysfunction.

The main aims of this study are to compare the limb coordination, balance, and gait profiles seen in adults with mitochondrial disease and people with a pure cerebellar syndrome secondary to spinocerebellar ataxia type 6 (SCA6), and to identify which features are sensitive to pathology.

At the end of this project, Dr Ng and his team (Dr Jane Newman and Dr Lia Alcock) hope to better understand which changes to gait are caused by degeneration of the cerebellum (as seen in SCA6), and which are caused by other co-existing symptoms of mitochondrial disease.

Lay Summary

Dr Ng and his colleagues are studying a group of conditions called mitochondrial disease. People with mitochondrial disease often have similar symptoms to people with ataxia, such as an unsteady gait and impaired balance, which is usually caused by dysfunction of the cerebellum.

When measuring balance and gait in people with mitochondrial disease, the researchers found that co-existing neurological symptoms, such as nerve damage and muscle weakness, can also cause problems with gait and balance. Dr Ng and his team want to know specifically which changes in gait are caused by degeneration of the cerebellum, and which are caused by other neurological symptoms. In order to better understand this, they plan to measure gait in people with SCA6. SCA6 is caused by degeneration of the cerebellum, and unlike mitochondrial disease it does not usually cause the other co-existing neurological symptoms.

Instruments used to measure gait are emerging as promising tools to measure and track the symptoms of ataxia. Developing sensitive ways of measuring the symptoms of ataxia is important for the design of clinical trials, to show whether a treatment in a trial improves the ataxia symptoms. As well as undergoing tests designed to measure gait and completing questionnaires at a hospital appointment, participants will also wear a small sensor on the lower back for seven days while at home. This will allow researchers to gather data on gait in people with SCA6 in their home environment, and compare this with measures taken at the hospital.

At the end of this project, Dr Ng and his team hope to better understand which changes to gait are caused by degeneration of the cerebellum (as seen in SCA6), and which are caused by other co-

existing symptoms of mitochondrial disease. The researchers expect that this project will also generate useful data in support of using gait measurements in future clinical trials.

For more support or information please contact: Ataxia UK, 12 Broadbent Close, London, N6 5JW
Website: www.ataxia.org.uk.
Helpline: 0800 995 6037 Tel: +44 (0)20 7582 1444
Email: helpline@ataxia.org.uk.