

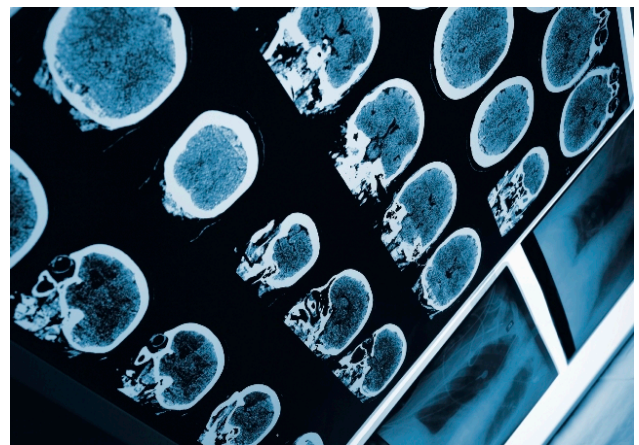


## The neurological basis of genetic diseases

**Professor Vaughan Macefield from the School of Medicine is collaborating with an international team\* of scientists to study a group of genetic diseases in order to understand their neurophysiological basis. The research has been funded by the National Health and Medical Research Council (NHMRC).**

'Hereditary Sensory and Autonomic Neuropathies (HSAN) are a group of neurological disorders that affect what we feel, how we move and the automatic functions of our bodies – somatosensory, sensorimotor and autonomic function', explains Professor Macefield. 'Five subtypes exist, with HSAN III the most common. The HSAN III subtype is associated with a lack of pain awareness, absent tendon reflexes and lack of muscle coordination when walking. HSAN IV is the second most common, coming with with non-existent pain and temperature perception, but non-affected reflexes and muscle coordination when walking. HSAN II is far less common and is associated with low tendon reflexes, loss of pain and thermal perception but no motor abnormalities.' This project aims to explain the neurophysiological basis of the sensorimotor and autonomic disturbances in HSAN II, III and IV by investigating the lack of muscle control and labile blood pressure in people with HSAN III, and investigate the ability of people with HSAN II, III and IV to regulate their body temperature and ability to detect painful stimulus.

Participants will undertake a range of tests designed to assess their ability to sense the position of their limbs, as well as their muscle control and balance. Heart and neural activity will be correlated using a combination of techniques. The body's autonomous response to warmed or cooled water will be analysed and the skin and core temperature recorded. Pain perception will also be tested in participants.



By studying this group of diseases our understanding of how normal physiology turns into disturbed physiology can grow. The outcomes of this research may include promising directions for future treatments of genetic disorders.

**Project Title:** Sensorimotor and autonomic dysregulation in hereditary sensory and autonomic neuropathies

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