

|                                 |
|---------------------------------|
| <b>Royal Melbourne Hospital</b> |
|---------------------------------|

**Ocular Motor Research Laboratory, Dept of Neurology, Royal Melbourne Hospital**

**Full Project Title:** Ocular motor abnormalities in subjects with fragile X mutation

Principal Researcher: A/Prof Owen B White

Associate Researchers: Dr Claire Meyniel, Dr Joanne Fielding, Ms Lynette Millist

**1. Introduction**

You are invited to take part in this research project. This is because you are the maternal grandfather of a patient with fragile X syndrome. The research project is aiming to investigate eye movements and their control by higher functions of the brain, in order to provide insights into the generation of the cognitive and movement disorders associated with abnormalities of the FMR1 gene. This pilot study may lead to a more extensive study which would be directed at defining information that might provide prognostic information in individuals with the varied forms of this genetic disorder.

Participation in this research is voluntary. If you don't wish to take part, you don't have to.

**2. What is the purpose of this research project?**

Little is known about the underlying mechanisms causing motor disorder in patients with disorders of the FMR1 gene. Studies of eye movement control provide a unique opportunity to examine higher order control and may provide diagnostic and prognostic information that will both permit differentiation of patients with varying neurological disorders, permitting appropriate therapy, but might also permit an insight to the outcome of the disorder in an individual.

Disorders of the FMR1 gene, in full expression produces a devastating neurological disorder, in some subjects, with both motor and intellectual disorder. Partial expression of the abnormality is associated with a wide variety of neurological disorders, including parkinsonism, tremor and imbalance, with a tendency to accelerated degeneration in some individuals.

Studies of eye movements permit a higher level of interpretation of motor control and insights into the mechanisms that result in the motor and cognitive disorders observed in patients. Abnormalities can be quantified in a way that can never be achieved in limbs.

If you decide you want to take part in the research project, please contact A/Prof Owen White or Dr Joanne Fielding through Royal Melbourne Hospital, Department of Neurology, on 03 9342 8448 and leave a message. We will contact you and provide further information.