HARVARD MEDICAL SCHOOL

MASSACHUSETTS GENERAL HOSPITAL

Information on Prion Disease Research Study

Study Title: **Biomarkers in Individuals at Risk for Prion Disease**

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What is the goal of this study?

Our goal is to lay the groundwork to test future prion disease therapeutics in people at risk for genetic prion disease -- **before they ever experience symptoms.**

Before misfolded, disease-causing prions ever form, the normal prion protein is present in the brain, in all of us. There is strong evidence that reducing levels of this normal prion protein with a drug, if such a drug were to be developed, would delay the onset of genetic prion disease.

There is also strong evidence that such a drug would be most effective if given to at-risk individuals before they experienced symptoms. But to test a drug in this group, **we need a way to measure whether it is having its intended effect, without relying on symptoms.** Therefore, we are gathering data on prion protein levels in cerebrospinal fluid (CSF). We already know that prion protein can be measured in CSF. Now we need to understand more about how stable it is over time in order to understand how well we could measure a drug-dependent reduction. And we need to gather data from the population that we would someday hope to treat.

Please note that this study is not a clinical trial. No one will be receiving an experimental drug in this study, and participation in this study is NOT a prerequisite to later participate in a clinical trial with an experimental drug. Your participation will contribute to science and help us prepare for clinical trials down the road, but there is no direct benefit to you for participating in this study, and participation is purely voluntary.

Who is eligible to participate?

You may be eligible for this study if any of the following apply to you:

- Have a family history of genetic prion disease, including genetic Creutzfeld-Jakob Disease (CJD), fatal familial insomnia (FFI), or Gerstmann-Straussler-Scheinker syndrome (GSS). Note: you do *not* need to know your genetic status to participate.
- Have received predictive testing indicating that you are at risk for one of the above diseases.

In addition, participants must be:

- Between the ages of 18 and 85
- Fluent in English

What does participation involve?

Potential volunteers first have a phone conversation with the study team to answer any questions, confirm eligibility, and collect some baseline information. For eligible individuals who decide to participate, the study involves annual visits to Massachusetts General Hospital in Boston, MA, with an interval between visits of 9-12 months.

The study visits would include the following components, performed over 1.5 days:

- Routine physical exam
- Health questionnaires
- Lumbar puncture
- Blood draw
- Brain imaging scans
- Cognitive tests
- Gait analysis

Eligible participants will receive compensation and financial assistance with travel costs. All participation is strictly voluntary.

Who should I contact to learn more or volunteer?

Contact MGHPrionStudy@partners.org