

## **DEVELOPMENT OF ASSAY FOR CREUTZFELDT-JAKOB DISEASE**

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This is a research study to develop a method for early diagnoses of Creutzfeldt-Jakob disease (CJD), Gerstmann-Sträussler syndrome, and other “prion” disorders using samples such as blood, saliva, urine or cerebral spinal fluid (CSF).

Currently, there is no test that can accurately diagnose these diseases without direct examination of brain tissue after death. We need to collect tissues from patients with and without the disease for use for our test.

This study is voluntary. If you decide to take part in this study, you can stop at any time.

This study is sponsored by the National Institutes of Health and National Institute of Neurological Disorders and Stroke.

This is a brief summary of what happens in this study:

1. Prior to study enrollment, you will undergo a comprehensive review of your medical history and records. You, your caregiver(s), and/or physician(s) will be asked questions about your current and past medical conditions, demographics, laboratory results, medications, family’s medical history
2. You will come to UCSF for additional evaluation and possible study enrollment. You might stay as an inpatient on the research floor of UCSF for 1 or 2 days.
3. At UCSF
  - You will undergo several diagnostic procedures or tests, some of which may be clinically necessary to confirm or rule out a diagnosis and others that, although clinically useful, are primarily for research purposes. These include, but are not limited to:
    - i. MRI (a scan that takes a picture of your brain)
    - ii. Electroencephalogram (EEG; measures brain waves)
    - iii. Lumbar puncture (spinal tap)
    - iv. Blood draw – about 200 ml (13 tablespoons) will be collected once and may be repeated a 2<sup>nd</sup> time
    - v. Urine sample
    - vi. Saliva sample
  - Genetic testing will be done on your blood sample to see if you carry the trait for a genetic prion disease. Counseling is

provided to help you decide if you would like to know the results of genetic testing.

- You and your caregivers will be asked questions about your medical history, the course of your illness, and your family medical history.
- You will have a brief medical examination, a videotaped neurological exam, and neurocognitive testing.
- You asked to return to UCSF, if possible, for follow-up visits.
- You will be asked to consent to an autopsy to establish a diagnosis and to provide tissues for research

The **risks** of this study are:

1. Discomfort, headache and infection (unlikely) from the lumbar puncture.
2. Stress, depression, hopelessness, fear of having children or fear that my current children will get the disease upon learning results of genetic testing.
3. Temporary discomfort from the blood draw, bruising, and rarely, infection.
4. Discomfort from lying still in the MRI machine or for the EEG.
5. Fatigue in undergoing neurological and neuropsychological testing.
6. Disclosure of personal medical information will result in a loss of privacy, however, records will be kept as confidential as possible.

The **benefits** of this study are:

1. There is no direct benefit to you by participating in this study.
2. Your participation may help in the early detection and treatment of future patients with prion diseases.

**For more information about this study, please contact:**

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