

Early Diagnosis of Human Prion Disease

Summary

- **Study director:** [Michael Geschwind, MD, PhD](#)
- **Sponsor:** National Institutes of Health
- **Recruiting?:** Yes
- **Official study title:** Early Diagnosis of Human Prion Disease
- **Conditions studied:** Transmissible spongiform encephalopathies and rapidly progressive dementias
- **Purpose:** The primary goal of this study is to develop and test methods for differentiating Creutzfeldt-Jakob disease (CJD) from other diseases and to learn how to diagnose CJD as early as possible. A secondary goal of this study is to learn more about how CJD and other rapid neurological conditions progress by studying these diseases in each patient over time. Because CJD is a rare condition and very rapidly progressive, diagnosis of the disease is often made very late in the illness. If any future treatments for CJD are to be successful, they likely will need to be given to patients as early as possible, before the disease progresses too far. It is therefore important that we learn how to make an accurate diagnosis earlier.

Eligibility

- **Inclusion criteria:** Patients must be 18 years of age or older. Must fulfill [WHO Revised Criteria for CJD](#) , have a family history of familial human prion disease, or have a history of progressive neurologic decline resulting in dementia or significant functional impairment from patient's baseline over two years or less from symptoms onset. Must have an informant who has frequent contact with the subject and is available to provide information about the subject.
- **Exclusion criteria:** Unable to undergo MRI due contraindication to this procedure (e.g., pacemaker, pregnant women). Known HIV positive or immunosuppressed patients.

What is involved?

- **Testing:** History and physical neurological examination, lumbar puncture, neuropsychological testing, MRI of the brain, EEG, MEG, nerve conduction study and blood draw. (For deceased and patients who cannot come to UCSF for a visit, we are only requesting the medical records for review and research purposes.)

- **Frequency of visits:** Will return for repeat MRI of the brain, EEG and MEG testing depending on the severity and progression of the specific neurologic conditions.
- **Materials needed prior to evaluation:** Diagnosis of a rapidly progressive dementia
- **Costs:** No costs will be charged for any of the study procedures.

Contact information

- **CJD/RPD Study Coordinator:** [Julie Thai](#) , 415-476-2909