

National Prion Disease Pathology Surveillance Center Testing and Reporting Policies

As a part of our surveillance efforts for CJD, the National Prion Disease Pathology Surveillance Center (NPDPS) conducts four different tests on the biopsy and autopsy samples we receive:

- ♦ Western blot: This test demonstrates the presence of the abnormal prion protein, which is believed to cause CJD and other prion diseases. If the abnormal protein is present, the case is positive. The Western blot is the most sensitive test for prion disease. This test is performed on frozen tissue.
- ♦ Immunohistochemistry (IHC)/Histology: In these tests, the neuropathologist examines slides of specially prepared brain tissue to see where the abnormal prion protein appears in order to help determine the type of prion disease. Different types of CJD have different distribution patterns of the abnormal protein. These tests are performed on fixed tissue.
- ♦ Genetic analysis: This test determines if the patient has a genetic mutation, and therefore a familial prion disease. The genetic analysis can only determine if a case is familial (which occurs in about 10% of positive cases); in all other forms of prion disease such as sporadic, iatrogenic, or variant CJD, the genetic analysis may help to identify the specific type. This test is performed on frozen tissue or blood. If we receive sufficient amounts of frozen tissue, blood is not required.

All four of these tests must be performed to provide a full diagnosis. We perform all of them providing that the appropriate samples are available. If one of the samples is not available, we cannot perform all of the tests and cannot provide a full diagnosis.

Although we perform all of the above tests for our important research efforts on prion disease, we realize that some families may not want all of the information we collect. In particular, some families do not want to receive genetic information. Genetic mutations not only affect the patient, but also other blood relatives who could also have the mutation. It is important to discuss the psychological implications, confidentiality and insurance with them to determine if they wish to receive this information.

In order to insure that the family receives only the information they want, we are asking clinicians to consult with families to determine if they would like to receive a full or partial diagnosis. Please indicate their choice on the attached form and fax it to us at **216-368-4090**. The NPDPS will not release genetic information until this form is returned.

For questions, please contact us at **216-368-0587** or cjdsurv@case.edu.

- Please send only a partial diagnosis, including the Western blot (if frozen tissue is available) and IHC/Histology (if fixed tissue is available), without the genetic analysis. The partial diagnosis will only tell if the case is positive or negative.
- Please send the full diagnosis, including the genetic analysis (only available if blood/frozen tissue is submitted). The full diagnosis will tell if the case is positive or negative and provide the type (sporadic and the subtype of sporadic, familial, or variant) of prion disease if the case is positive.

Patient Name: _____

Physician Signature: _____

Date: _____