Brain biopsy for Creutzfeldt-Jakob Disease Diagnosis

The current diagnostic criteria are limited; test sensitivity and specificity vary with the genetics of the disease as well as the clinical stage. Physicians may be unsure of all diagnostic testing available and may order outdated tests or prematurely request a brain biopsy when the diagnostic workup is incomplete [1)].

Heinemann et al. studied the records on 26 brain biopsies conducted in patients with suspected Creutzfeldt-Jakob Disease who had been referred to the CJD Surveillance Unit in Germany between 1993 and 2005.

Of the 26 included patients, 11 suffered from neuropathologically confirmed CJD, which in 5 cases had been deemed clinically “probable” and in 2 had been deemed “possible.” The disease in the remaining 4 patients had been categorized as “other” prior to neuropathological confirmation of CJD. The results of 15 brain biopsies showed no features of prion disease. None of these 15 patients had received a probable diagnosis of CJD, 4 had a possible diagnosis, and 11 had received a diagnosis of “other.” Three of the cases classified as other and none of those with CJD presented with pleocytosis in the CSF. In 73% of the other cases, biopsy sampling did not reveal any results characteristic of CJD but did not provide specific findings on which to base a differential diagnosis. An autopsy confirmed the biopsy diagnosis of CJD in all cases and additionally confirmed that CJD was not present in 3 patients who had nondiagnostic biopsy results.

Biopsy sampling may be helpful in the diagnostic approach to rare cases of dementia for which a reliable diagnosis cannot be established on the basis of clinical symptoms, CSF parameters, electroencephalography, and MR imaging results [2)].

Due to lack of effective treatment and the potential for iatrogenic infection in surgery, a brain biopsy is reserved for cases where establishing the Creutzfeldt-Jakob Disease Diagnosis is deemed important, or as part of a research study, [3)] or when diagnostic tests are equivocal and other potentially treatable etiologies are suspected.

To prevent aerosolization of the infectious agent, a manual saw is recommended over a power craniotome, and every effort should be made to avoid cutting the dura with the saw. Recommended decontamination procedures should be followed

Specimens should be clearly labeled as being from suspected CJD patients to alert laboratory personnel to the hazard. Tissue should be fixed in a saturated 15% phenolized formalin (15 g of phenol per dl of 10% neutral buffered formalin with the undissolved phenol layering at the bottom of the solution) [4)].

Analysis for classic histologic findings and/or immunostaining for PrPres are the gold standards of diagnosis.

1)

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2)

Heinemann U, Krasnianski A, Meissner B, Kallenberg K, Kretzschmar HA, Schulz-Schaeffer W, Zerr I. Brain biopsy in patients with suspected Creutzfeldt-Jakob disease. J Neurosurg. 2008 Oct;109(4):735-41. doi: 10.3171/JNS/2008/109/10/0735. PMID: 18826363.
3)

Schlitt MJ, Morawetz RB, Bonnin JM, et al. Brain Biopsy for Encephalitis. Clin Neurosurg. 1986; 33: 591–602
4)

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