Creutzfeldt-Jakob Disease Diagnosis

Diagnostic criteria

The complete “diagnostic triad” (dementia, myoclonus, and periodic EEG activity) may be absent in up to 25% of cases of Creutzfeldt-Jakob Disease. Diagnostic criteria have been published [1] No patients in their series with a diagnosis other than CJD fulfilled the criteria for clinically definite CJD. The most common condition other than CJD fulfilling the criteria for clinically probable CJD was senile dementia of the Alzheimer type (especially difficult to distinguish in the early stages).

In patients with dementia, a positive immunoassay for the 14-3-3 protein in cerebrospinal fluid strongly supports a diagnosis of Creutzfeldt-Jakob disease. This finding, however, does not support the use of the test in patients without clinically evident dementia [2]

Diagnostic tests

Imaging

No characteristic CT or MR finding. These studies are frequently normal but are essential to rule out other conditions, (e.g. herpes simplex encephalitis, recent stroke...). Diffuse atrophy may be present, especially late. MRI may show increased intensity on T2 weighted image in areas typically involved (basal ganglion, striatum) in up to 79% of cases (retrospectively) [3]. This is nonspecific but may help differentiate CJD from senile dementia of the Alzheimer type [4].

Blood tests

Serum assays for S-100 protein are so insensitive and nonspecific that it can only be used as a diagnostic adjunct

CSF

Cerebrospinal fluid analysis for Creutzfeldt-Jakob Disease Diagnosis.

EEG

Characteristic finding of bilateral, symmetrical, periodic bi- or triphasic synchronous sharpwave complexes, AKA periodic spikes, AKA pseudoperiodic sharp-wave complexes (0.5–2 per second) have ≈ 70% sensitivity and 86% specificity. They resemble PLEDs, but are responsive to noxious stimulus (may be absent in familial CJD19 and in the recent UK variant

SPECT scan

May be abnormal in vCJD even when EEG is normal36; however, the findings are not specific for vCJD.

Tonsillar biopsy

Patients with variant CJD (vCJD) may have detectable levels of variant type 4 of the abnormal prion protein (PrPSc) in their lymphoreticular system, which may be accessed by a 1cm wedge-biopsy of one palatine tonsil (using careful aseptic precautions)

Brain biopsy

Brain biopsy for Creutzfeldt-Jakob Disease Diagnosis.

1)
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Hsich G, Kenney K, Gibbs CJ, Lee KH, Harrington MG. The 14-3-3 brain protein in cerebrospinal fluid as a marker for transmissible spongiform encephalopathies. N Engl J Med. 1996 Sep 26;335(13):924-30. doi: 10.1056/NEJM199609263351303. PMID: 8782499.
3)
Finkenstaedt M, Szudra A, Zerr I, et al. MR Imaging of Creutzfeldt-Jakob Disease. Radiology. 1996; 199: 793–798
4)
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