Pre-mortem Diagnosis of Sporadic Creutzfeldt-Jakob Disease in Practice

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Abstract

We describe a case of Sporadic Creutzfeldt-Jakob disease (sCJD) and discuss our evidence-based diagnostic process.

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Definite Sporadic Creutzfeldt-Jakob Disease

Diagnosed by standard neuropathological techniques; and/or immunocytochemically; and/or Western blot confirmed protease-resistant PrP; and /or presence of scrapie-associated fibrils.

Probable Sporadic Creutzfeldt-Jakob Disease

Neuropsychiatric disorder plus positive RT-QuIC in cerebrospinal fluid (CSF) or other tissues

OR

Rapidly progressive dementia; and at least two out of the following four clinical features:

1. Myoclonus
2. Visual or cerebellar signs
3. Pyramidal/extrapyramidal signs
4. Akinetic mutism

AND a positive result on at least one of the following laboratory tests

- a typical EEG (periodic sharp wave complexes) during an illness of any duration
- a positive 14-3-3 CSF assay in patients with a disease duration of less than 2 years
- High signal in caudate/putamen on magnetic resonance imaging (MRI) brain scan or at least two cortical regions (temporal, parietal, occipital) either on diffusion-weighted imaging (DWI) or fluid attenuated inversion recovery (FLAIR)

AND without routine investigations indicating an alternative diagnosis.

Possible Creutzfeldt-Jakob Disease

Progressive dementia; and at least two out of the following four clinical features:

1. Myoclonus
2. Visual or cerebellar signs
3. Pyramidal/extrapyramidal signs
4. Akinetic mutism

AND absence of positive result for any of the 4 tests above that would classify a case as "probable"

AND duration of illness less than two years

AND without routine investigations indicating an alternative diagnosis.