Creutzfeldt-Jakob Disease (CJD)

Dementia Neuropsychology · https://neuro.vip/neuropsychology/dementia-neuropsychology/creutzfeldt-jakob-disease-cjd/

Creutzfeldt-Jakob Disease (CJD) is a rare, rapidly progressive neurodegenerative disorder caused by **misfolded prion proteins**. It is particularly significant within clinical neuropsychology due to its **swift cognitive decline, distinctive symptom profile, and challenging differential diagnosis**. Early recognition is vital given its prognosis, impact on patients and families, and the complexities of management and care planning.

Creutzfeldt-Jakob Disease Nature and Aetiology

CJD belongs to the group of human prion diseases, alongside variant CJD, fatal familial insomnia, and Gerstmann-Sträussler-Scheinker syndrome. It may arise sporadically (most common), be inherited due to mutations in the PRNP gene, or result from exposure to infectious prion-containing tissues (iatrogenic or variant forms).

Pathologically, CJD is characterised by **spongiform changes in the brain, neuronal loss, gliosis, and the accumulation of abnormal prion proteins**. This underpins its **strikingly rapid and devastating clinical course**, distinguishing it from most other dementia syndromes.

Core Clinical Features in Creutzfeldt-Jakob Disease

Cognitive Profile

CJD presents with a rapidly progressive dementia—most patients advance from mild symptoms to profound cognitive dysfunction within weeks or a few months. Early presentations may include:

- Impaired **memory and attention** (often with marked working memory decline)
- **Disorientation** and reduced insight
- **Language** disturbances (reduced fluency, word-finding difficulty, comprehension problems)
- Perseveration and impaired **executive functioning**

Neurobehavioural and Psychiatric Changes

Mood disturbance, anxiety, irritability, and apathy are common, and frank psychosis can occur in the early stages. Visual hallucinations may also be reported.

Neurological Signs

CJD's hallmark is its mix of neuropsychiatric and neurological features. Neurological signs often emerge during the illness and may include:

- Myoclonus (involuntary muscle jerks), often provoked by sudden movement or noise
- Ataxia, unsteadiness, and gait disturbance
- Dysarthria, dysphagia, and other signs of widespread cortical and subcortical dysfunction
- Visual disturbances (e.g., cortical blindness or field defects)

As the disease progresses, patients become akinetic, mute, and rigid, with a typical disease duration of less than a year (often only several months).

Brain Imaging in Creutzfeldt-Jakob Disease

MRI brain, EEG (often showing periodic sharp wave complexes in sporadic CJD), CSF tests (including RT-QuIC for prion protein), and blood tests are vital adjuncts, but the neuropsychological profile is a key part of the clinical synthesis.

Embedded media: https://radiopaedia.org/cases/45562/play?lang=gb

PET, CT and MRI imaging of a real CJD case in a 55-year old with "diagnosis certain" © Radiopaedia

Role of the Clinical Neuropsychologist in Creutzfeldt-Jakob Disease

The clinical neuropsychologist's expertise is crucial in:

- Early detection of the specific cognitive and behavioural features
- Supporting differential diagnosis through neuropsychological profiling
- Educating family members and the clinical team about the illness trajectory and care needs

- Contributing to capacity assessments, care planning, and providing input to palliative care discussions

Comprehensive assessment may be affected by rapidly declining function, reduced cooperation, and fluctuating levels of awareness, requiring flexibility in approach.

Differential Diagnosis in Creutzfeldt-Jakob Disease

CJD should be distinguished from other causes of rapidly progressive dementia and other neuropsychiatric syndromes. Key differentials include:

- **Rapidly Progressive Alzheimer's Disease:** May mimic CJD but typically lacks the prominent myoclonus and neurological signs.
- **Autoimmune / Paraneoplastic Encephalopathies:** These may present with neuropsychiatric features and encephalopathy but often have distinctive antibodies and may respond to immunotherapy.
- **Viral or Infectious Encephalitis:** Can cause acute cognitive and neurological symptoms, sometimes with fever and seizures.
- **Vascular Dementia (multi-infarct):** Sporadic step-wise decline with focal neurological findings, but usually a more protracted course.
- **Other Prion Diseases:** Such as variant CJD, which tends to affect younger individuals and may be associated with prominent psychiatric symptoms at onset.
- **Toxic, Metabolic, and Neoplastic Encephalopathies:** Consider if systemic or metabolic signs are present.

Points for Clinical Practice

- **Speed of Decline:** The rapidity of cognitive and functional deterioration is a key distinguishing feature, with a timeline measured in months rather than years.
- **Multifaceted Presentation:** CJD blends psychiatric, cognitive, and neurological symptoms, and the constellation—particularly myoclonus and ataxia alongside dementia—is highly suggestive.
- **Neuropsychological Assessment Approach:** Serial brief cognitive assessments may track the profound, rapid decline more effectively than full batteries. Bedside assessments of cognition and behaviour can be particularly helpful.

Summary

CJD is a rare but essential diagnosis to consider in rapidly progressive dementias. Neuropsychologists play a crucial role in detecting its distinctive cognitive and neuropsychiatric features, supporting differential diagnosis, and advising teams and families about its swift course and significant support needs. Differentiating CJD from other treatable causes of rapidly progressive dementia is vital, making expert neuropsychological assessment a key part of the multidisciplinary approach.