Early involvement of cranial nerves in a patient affected by E200K familial Creutzfeldt-Jakob disease

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Creutzfeldt-Jakob disease (CJD) is a rare, sporadic, familial or iatrogenic human prion disease, characterized by progressive dementia and different neurological symptoms and signs.

Early involvement of cranial nerves as initial symptom of CJD is uncommon.

The current report describes an elderly man with E200K familial CJD showing a prominent involvement of cranial nerves.

77-year-old man

previous metastatic colic cancer in 2014

- on Jenuary 2016, the patient developed subacute dysarthria associated with decreased alertness

- on admission to our neurology ward, the neurological examination showed cranial multineuropathy, characterized by

- right eyelid ptosis
- horizontal gaze palsy
- hypophonia
- dysarthria
- moderate dysphagia

A left pyramido-cerebellar syndrome was also noted

Although the mild decreased consciousness, a bedside MMSE was within normal range

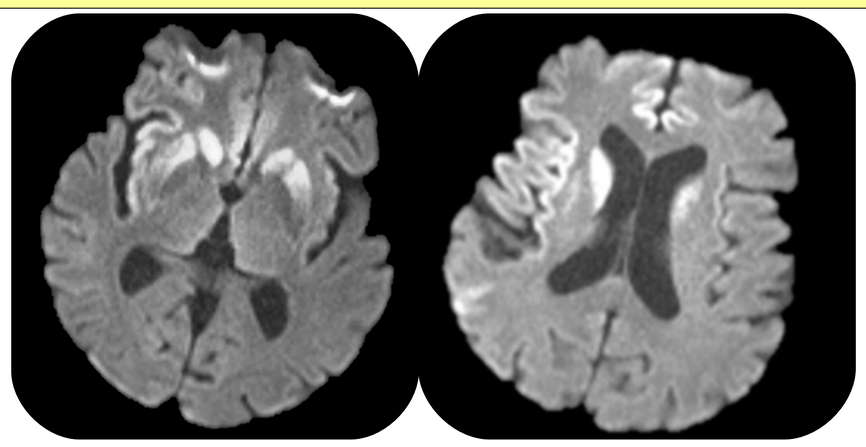
Differential Diagnosis (I)

- **cranial multineuropathy due to meningeal carcinomatosis**
- **paraneoplastic syndrome with prevalent involvement of cranial nerves**

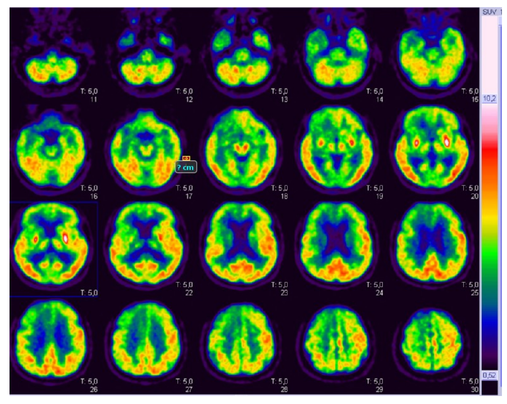
Diagnostic work-up

- **EEG:** right hemispheric and left anterior slowing
- **CSF examination:** unremarkable

- **Brain MRI: restricted diffusion areas on DWI sequences at basal ganglia, paramedian frontal cortex bilaterally, right temporo-insular cortex**

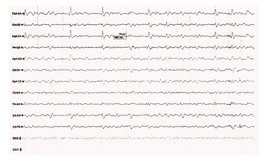


- **Brain PET: cortical hypometabolism associated with bilateral putaminal hypermatabolism**



Differential Diagnosis (II)

- **paraneoplastic neurological syndrome**
  (considering the atypical clinical features)
  - **I cycle of IGIV (0,4mg/kg/die for 5 days), without any clinical response**
- **CREUTZFELDT JACOB DISEASE**
  - **rapid worsening of cognitive performances (MMSE 8/30)**
    mutacic state with upper limbs dystonic movements associated to oro-buccal diskynesia



- **SERIAL EEG finally revealed PERIODIC WAVES**
- **14.3.3 PROTEIN detection ON CSF was POSITIVE**

The genetic test was performed, revealing a E200K mutation in the PRNP gene associated to the MM polymorphism on codon 129

The patient died one month later, due to nosocomial pneumonia

Our case confirms the already known wide phenotypic clinical variability of patients with fCJD and widens the clinical spectrum of the disease

In line with recent literature FLAIR and DWI abnormalities at basal ganglia and cortex offer best sensitivity and specificity for early diagnosing fCJD, as for sporadic CJD

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Creutzfeldt-Jacob disease with E200K PRNP mutation: a case report and revision of the literature, Murri et al., 2009
Prion disease genetics, Mead, 2006