Diagnostic tools through different clinical stages of Creutzfeldt-Jakob Disease: a case report.

Tappatà M, Introna A, Chiapparino C, Francavilla T, Leante R, Dicuonzo F, Simone IL and Tortorella C.

Department of Basic Medical Sciences, Neurosciences and Sense Organs, University of Bari

INTRODUCTION: Creutzfeldt-Jakob Disease (CJD) is a spongiform encephalopathy leading to rapidly progressive dementia (RPD). It could be differentiated from other forms of RPD ante-mortem by the detection of specific clinical signs associated with specific paraclinical findings, such as periodic sharp wave complexes on EEG, detection of 14-3-3 protein in cerebrospinal fluid (CSF) and hyperintensity of cortical regions or caudate and striatum nuclei. Recently, the role of CSF p-tau/tau ratio has been considered challenging for CJD diagnosis.These markers might show different sensibility in relation to symptoms progressing.

CASE REPORT: A 57 years old caucasian woman was hospitalized for postural instability developed over two months. At neurological examination the patient showed cognitive impairment, memory loss with anosognosia, severe ataxia, fragmented ocular saccades, myoclonus, dysmetria and dysarthria. Brain MRI revealed DWI and FLAIR hyperintensity in temporal-parietal-occipital cortex, both talami and basal ganglia (Figure 1). CSF analysis showed low p-tau/tau ratio (0.013), but no detection of 14-3-3 protein. On EEG isolated sharp bi-triphasic waves were recorded on temporal-parietal- occipital regions (Figure 2). Standard autoimmunity and antibodies for viral encephalitis were negative. Genetic test was negative for PrP mutation, with Met-Val polymorphism. We then made a diagnosis of sporadic Probable CJD, even if MRI was the only paraclinical data supporting diagnostic criteria. Two months later the patient worsened with a complete loss of her home independence. Cerebellar and cognitive symptoms worsened too, and diffuse hypertonia became evident. Brain MRI pointed out strengthened cortical DWI hyperintensity and new cortical ribbons of the frontal cortex (Figure 3).
A second CSF analysis was performed; 14-3-3 was now detected and further reduction of p-tau/tau ratio (0.010) was found. EEG was slowed with diffuse presence of theta sharp waves in temporal-parietal-occipital regions even if periodism was still not clear (Figure 4).
The diagnosis of Probable CJDs was confirmed.

FIGURE 1

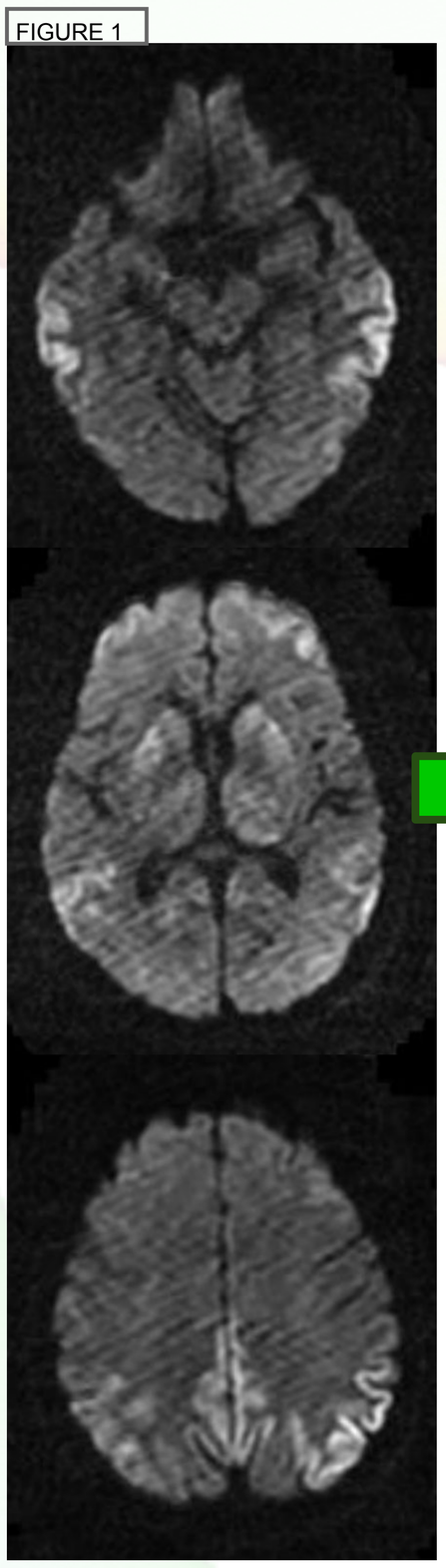


FIGURE 3

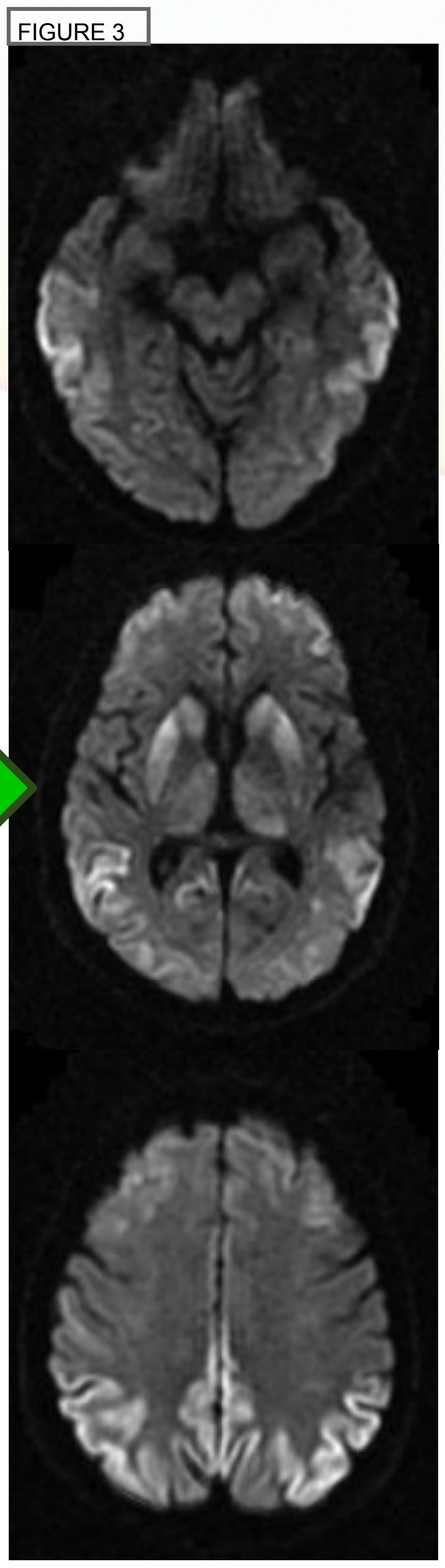


FIGURE 2

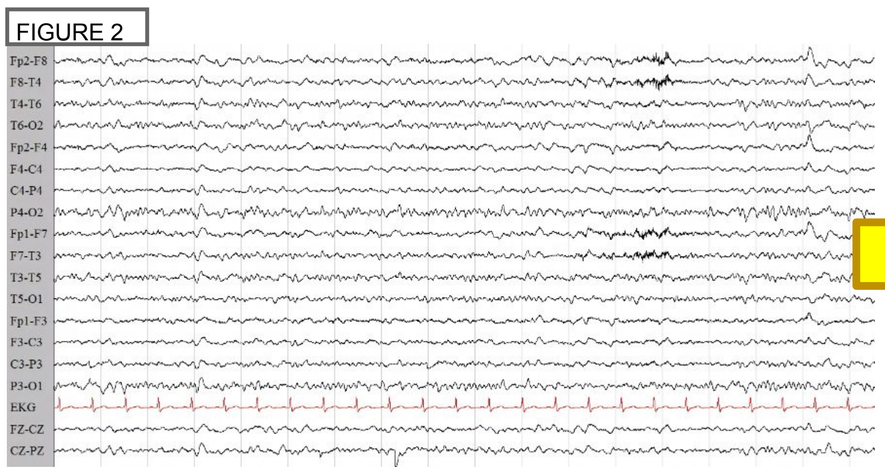
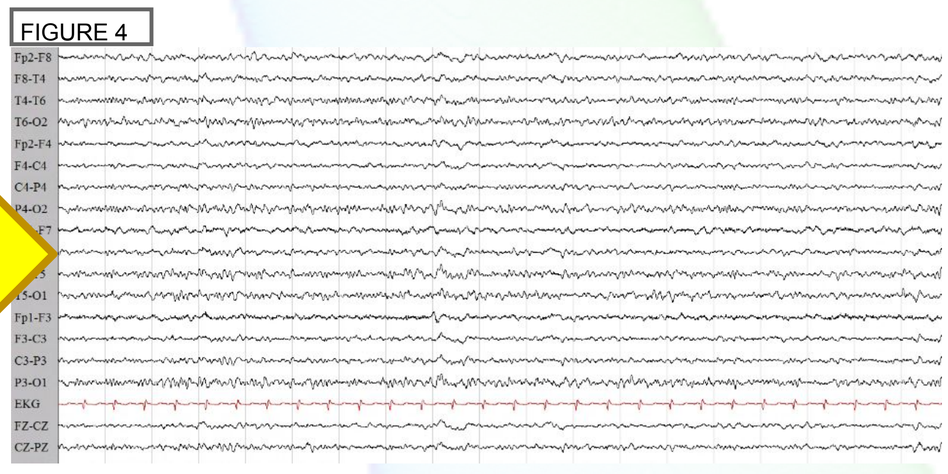
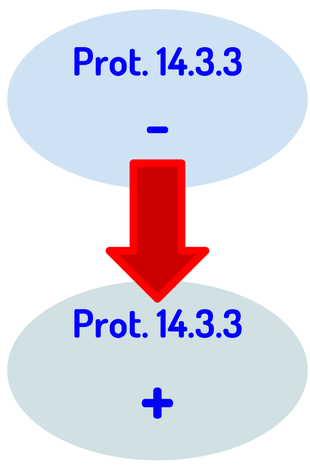


FIGURE 4





CONCLUSIONS: Diagnosis of CJD could be difficult in the early stages when clinical signs might not be supported by instrumental and biochemical findings. In our case MRI abnormalities were confirmed to be the first and more sensitive tool for early diagnosis, while 14-3-3 CSF protein and EEG findings seem to take more time to reveal. Moreover, even if p-tau/tau ratio is not yet included in the diagnostic criteria, our data support the evidence of a key role for the diagnosis since the early stages.

References:
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