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A Case of rapidly progressive sporadic Creutzfeldt-Jakob Disease

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Abstract
Creutzfeldt-Jakob disease (CJD) is a rare fatal rapidly progressive dementing disorder. The estimated annual incidence was 0.5 to 1.5 million populations. The illness is relentlessly progressive and generally causes death within 9 months of onset. Most CJD patients presents between 60 and 75 years of age.
Here by we report a case of sporadic Creutzfeldt-Jakob disease presenting as rapidly progressive neurodegenerative disorder.
Keywords: *Creutzfeldt-Jakob, Prion, Delta activity.*

Introduction

Creutzfeldt - Jakob disease (CJD) is a rare fatal rapidly progressive dementing disorder. Early symptoms include memory problems, behavioural changes, poor coordination, and visual disturbances. It was first described in the year 1920. The estimated annual incidence was 0.5 to 1.5 million populations. The illness is relentlessly progressive and generally causes death within 9 months of onset. Most CJD patients presents between 60 and 75 years of age.

Here by we report a case of sporadic Creutzfeldt-Jakob disease presenting as rapidly progressive neurodegenerative disorder.

Case Report

A 49-year-old lady who is an alcoholic vendor by occupation presented with asthenia for 2 months, right upper limb dystonia and slowness in daily activities for 1-1/2 month. She had memory and speech disturbances and loss of comprehension for 20 days. She had Chorea for 15 days and myoclonic jerks for 4 days prior to presentation to hospital. On examination, patient had axial and limb rigidity, all deep tendon reflexes are brisk and limb ataxia present.

Investigations

Diagnosis involves ruling out other potential causes.
Routine blood investigations are within normal limits
CSF analysis shows TC-5 cells/mm, DC- 100% lymphocytes, Protein-18mg/dl, Sugar-74mg/dl, ADA- Negative.
Fungal stain, Gram stain, Bacterial culture are Negative.
TPO antibodies -12.8 IU/ml.
MRI brain shows cortical ribboning prominent at occipital cortex.
EEG shows diffuse slowing with delta activity.

a) 14-3-3 protein dot blot

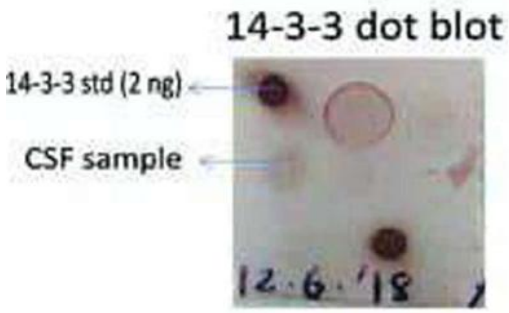

Fig. 1 CSF 14-3-3 protein dot blot

b) MRI Brain

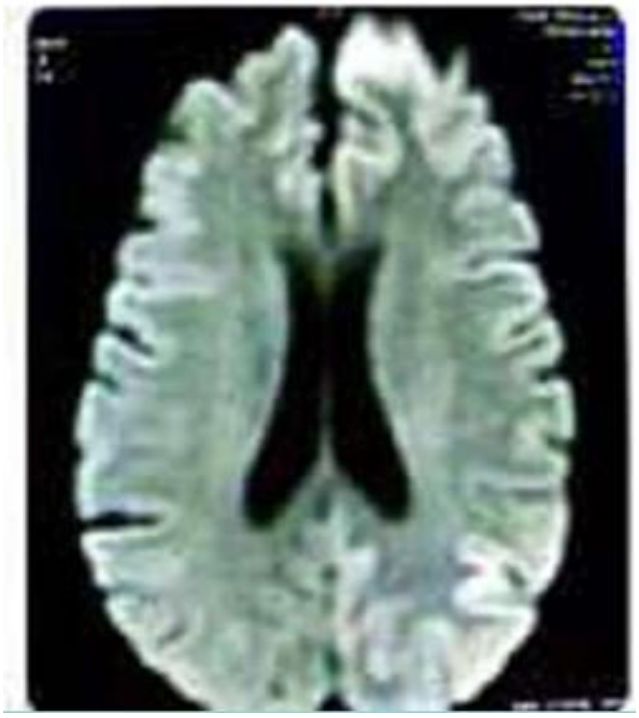

Fig. 2 DW MRI image

c) EEG

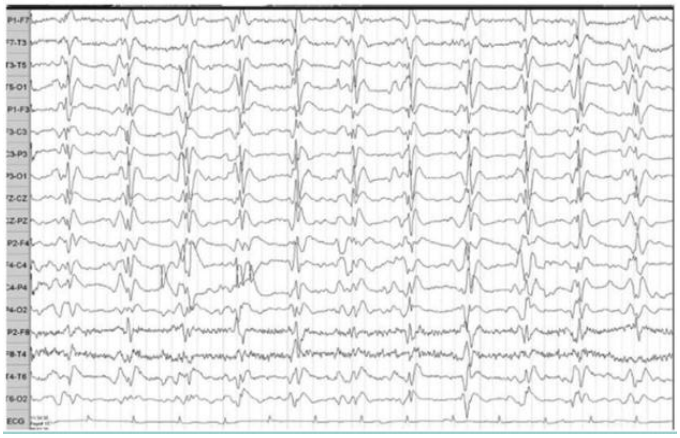

Fig. 3 EEG showing delta activity

Discussion

CJD first described by Hans Gerhard Creutzfeldt in 1920 caused by misfolded prion protein that affect signalling processes, damaging neurons and resulting in degeneration.

Sporadic CJD is the most common prion disease which is under reported in India. NIMHANS reported 69 cases in 30 years (1968-1997) and in 2016, 15 cases of CJD were collected over 4 years. In North India 10 cases of CJD were reported from 1990 to 1998. In Kerala, a tertiary care hospital reported 30 cases of probable CJD from 2000 to 2013. We report a case of CJD from Government tertiary care hospital, Kakinada admitted in 2018 with age of presentation before 50 years.

The case ended in akinetic mute state rapidly irrespective of initial manifestations. The main diagnostic tools were MRI Brain DW imaging, CSF 14-3-3 and EEG as in other studies.

Conclusion

CJD is a rapidly progressive and highly heterogenous condition that readily mimics number of conditions which may be reversible.

It rapidly progresses to mute akinetic state associated with myoclonus universally irrespective of manifestation present initially. Greater awareness and high degree of suspicion only can give us the true scenario of this dreaded disease, for which no treatment is available till date.

Highly sensitive and specific testing as diffusion weighted and CSF RT-QuIC (quaking induced conversion) can confirm the diagnosis and lead to early hospital care for this universal fatal disease and open drug development for treatment.

There is no specific treatment. Opioids may be used to help with pain, while clonazepam or sodium valproate may help with involuntary movements.

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