

CREUTZFELDT-JAKOB DISEASE (CJD)

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In this article we present two cases of Creutzfeldt-Jakob Disease (CJD) diagnosed on the basis of rapidly deteriorating dementia, myoclonus and periodic triphasic waves on electroencephalogram (EEG).

CASE 1

A 75-year-old lady was admitted with a four week history of depression, slowed mobility and personality change. She then developed disturbance of gait and odd behaviour. There was no past history of cerebro-vascular or psychiatric disease. She lived alone and managed herself reasonably well and quite independently.

On examination, she was conscious but appeared depressed and generally slowed. Vital signs were normal. Central nervous system examination revealed that she had marked poverty of speech. Her gait was ataxic. Her Abbreviated Mental Test score was 2/10. There was no focal neurological deficit. Initial investigations including a full blood count, biochemical and thyroid profiles were normal. Normal renal and liver profile ruled out metabolic encephalopathy. A CT scan of the brain was also normal. She was transferred to the psychiatric hospital for treatment of depression. She was later noted to have prominent auditory hallucinations and myoclonus of limbs. An EEG was performed which showed triphasic waves at 1/sec interval and a diagnosis of CJD was made (Fig 1). She continued to deteriorate rapidly and within two weeks of admission became fully dependent on extensive nursing care. She died within six months of initial presentation.

CASE 2

A 76-year-old lady was admitted with a six week history of incoherence, problems with speech and inability to

communicate. She was having multiple falls and became incontinent of urine. She showed abnormal posturing from time to time and lost the ability to dress and toilet within days of admission. She then developed myoclonus of upper and lower limbs. Prior to the onset of illness she lived a fairly independent life.

Examination revealed that she was fully conscious, but unable to communicate verbally. She had had episodes of drowsiness. There was marked abnormal rigid posturing and myoclonus was noted in the limbs. Investigations showed a normal full blood count and biochemical profile including normal renal and hepatic profile. Examination of cerebrospinal fluid was also normal. A CT scan of the brain showed generalised involutinal changes. An MR scan was performed but proved inconclusive. An EEG showed periodic triphasic waves characteristic of Creutzfeldt-Jakob disease in the absence of a metabolic cause.

Her course in hospital was marked by progressive deterioration over the next couple of weeks and she became completely bed-ridden. She needed full nursing care and was later transferred to the hospice. She died in the hospice within four months of the onset of illness.

DISCUSSION

CJD is a rare but invariably fatal degenerative disease of the central nervous system. It is found worldwide with an annual incidence of 1 per million. Eighty percent of cases are sporadic and in fifteen percent of cases it may be inherited.

Although its exact aetiology is yet to be determined, it is widely regarded as a 'Human Prion Disease'. Prions are small proteinaceous infectious particles (not viruses) which resist inactivation by procedures that modify nucleic acids.

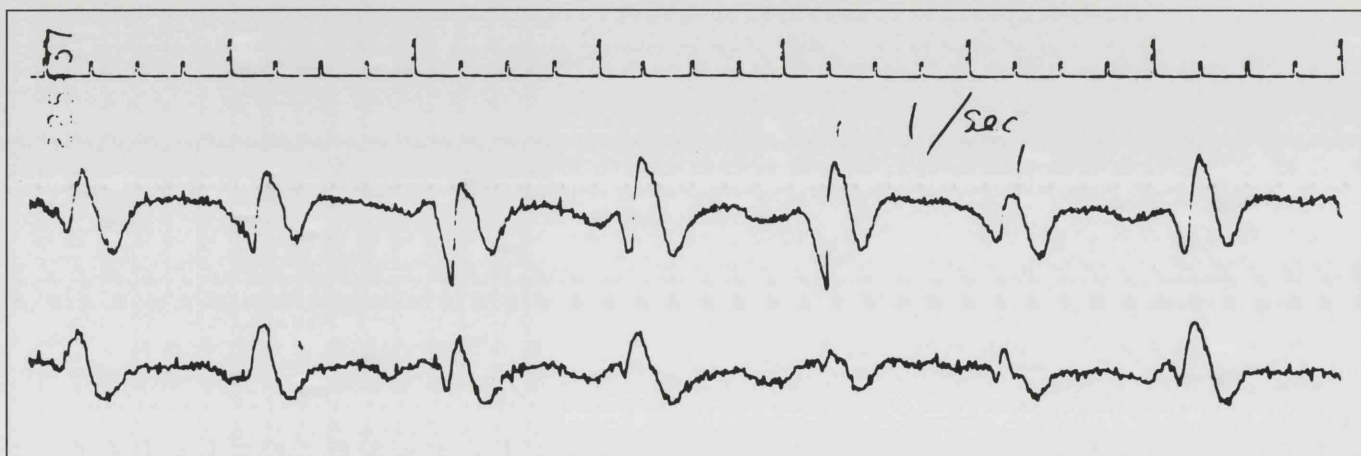


Fig 1 – Electroencephalogram showing periodic triphasic waves (1/sec) in CJD.

The major feature which distinguishes prions from viruses is the finding that prion protein is encoded by a chromosomal gene. In humans the prion protein gene is located on the short arm of chromosome 20. Prion is unique in being both inheritable and transmissible. Homozygous prion protein genotype predisposes to sporadic CJD¹. More recently, it has been found that the E⁴ allele of Apolipoprotein E is a risk factor for CJD². Person-to-person transmission has been noted to occur following treatment with cadaveric human growth hormone, corneal transplantation and use of contaminated intracerebral electrodes. Neuropathologically, CJD is characterized by spongiform degeneration of the brain (therefore also known as spongiform encephalopathy) and astrogliosis with a lack of inflammatory response³.

The clinical picture is dominated by rapidly progressive dementia and myoclonus. There is usually disturbance of gait, stance and motor control. Corticospinal and extrapyramidal signs may be seen. Bizarre behaviour and hallucinations are sometimes striking features.

CJD is confirmed on the basis of neuropathological findings. In the absence of neuropathological proof, a probable diagnosis of CJD can be made in a case of rapidly progressive dementia with characteristic EEG findings and at

least one of the following features: myoclonus, pyramidal signs, cerebellar signs, extrapyramidal signs.

The course of the disease is uniformly and progressively downhill right from the onset and death usually occurs in three to 12 months.

REFERENCES

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