INTRODUCTION

CJD is a rare, fatal neurological disorder that is caused by infectious proteinaceous particles called prions. Sporadic (sCJD), familial (fCJD), iatrogenic (iCJD), and variant forms of CJD (vCJD) are all recognized. The vast majority of CJD cases are sporadic (85 – 95%), while 5 – 15% are due to fCJD; iCJD generally accounts for less than 1%. CJD is characterized by rapidly progressive mental deterioration and myoclonus. Mental deterioration may manifest as dementia, behavioural abnormalities, and deficits involving higher cortical function. Myoclonus, especially provoked by startle, is present in more than 90% of patients at some point during the illness. Other features such as seizures, visual and speech disturbances, vertigo, ataxia and psychiatric manifestations have also been described. Till date, only one case of CJD has been reported from Pakistan.2 Here, we report a case of sCJD.

CASE REPORT

A 50 years male was admitted in Military Hospital, Rawalpindi, with 6 months history of progressive memory impairment and cognitive decline. These symptoms were preceded by disordered sleep, decreased appetite and easy fatigability. He was unable to concentrate properly and perform simple mathematical skills. Prior to his illness, he was recognized to be exceptionally good with numbers, meticulous and organized. His family members noticed that he remained generally in low mood and depressed. Later on, he started having difficulty in recalling past-events and recognizing his relatives. He lost interest in day-to-day routine activities and became socially withdrawn. Four months later, patient developed vertigo and unsteady gait to the extent that he was unable to walk without support. He started having myoclonic jerks, more in upper limbs than lower. Two weeks before presentation, he developed generalized seizures with urinary and bowel incontinence. He became progressively more sedentary and finally bed ridden and mute. During the course of disease, the patient had been visiting various physicians but his symptoms did not improve. There was no past history of neuropsychiatric disorder or any surgical intervention. He was non-addict. His family history was not significant.

General physical examination revealed an ill-looking gentleman lying in bed, awake but mute, and unaware of his surroundings. Other features such as seizures, visual and speech disturbances, vertigo, ataxia and psychiatric manifestations have also been described. Till date, only one case of CJD has been reported from Pakistan.2 Here, we report a case of sCJD.

ABSTRACT

A case of 50 years of age, male with sporadic Creutzfeldt Jakob Disease (sCJD) is reported. Patient had dementia, behavioural abnormalities, unsteady gait and myoclonic jerks. Magnetic resonance imaging (MRI) brain T2 weighted and Fluid Attenuated Inverse Recovery (FLAIR) images showed abnormally increased signal intensity in caudate nucleus and putamen. Scalp electroencephalogram (EEG) revealed periodic synchronous biphasic sharp wave complexes. On the basis of history, clinical findings, typical MRI brain and EEG changes, diagnosis of sporadic CJD was made.

Key words: Creutzfeldt Jakob disease. Dementia. Myoclonus.
showed abnormally increased signal intensity in caudate and putamen (Figure 1). EEG revealed periodic synchronous biphasic sharp wave complexes (Figure 2). Patient was diagnosed as a case of sCJD, fulfilling World Health Organization criteria for probable sCJD.3 Brain biopsy was required for definite diagnosis for which family members did not consent. Patient was given clonazepam and valproic acid to control seizures and myoclonus, and was provided nursing care. Patient's condition kept on deteriorating and he passed away on 13th day of admission.

**DISCUSSION**

The first case of CJD was documented in 1920 by two German doctors, Hans Gerhard Creutzfeldt and Alfons Maria Jakob. Approximately one case of sCJD occurs per 1 million population per year with a worldwide distribution. The mean age for the onset of disease is between 57 and 62 years, although rare cases in young adults and those over 80 years of age have been described.4 Some clinical findings, although compatible with CJD, should raise the suspicion of an alternative diagnosis, especially if they are among the more prominent features of the illness. These include cranial nerve abnormalities, sensory abnormalities, involvement of the peripheral nervous system, disturbances of pupillary responses and extracocular movements.

A characteristic EEG pattern of periodic synchronous bi- or triphasic sharp wave complexes (PSWC) is observed in 67 – 95% of patients with sCJD at some time during the course of the illness. Objective diagnostic EEG criteria proposed in 1996 was found to have a sensitivity and specificity of 67 and 86 percent, respectively for the diagnosis of CJD.5 MRI is not a criterion for the diagnosis of sCJD and it is not included in current WHO diagnostic criteria, although typical changes have been described in literature. Abnormally increased signal intensity in the putamen and head of the caudate in T2 weighted and FLAIR image is the most common finding on conventional MRI sequences in patients with CJD.6 Newer MRI sequences have been shown to be useful in diagnosing early lesions in CJD. Proton density and diffusion weighted image (DWI) are more sensitive than T2-weighted and FLAIR images for the detection of CJD specially in early stages of disease, showing abnormalities before the onset of characteristic clinical findings such as myoclonus and periodic sharp wave complexes seen on EEG.7 However, these findings are not entirely specific for CJD and may be confused with stroke, vasculitis, or reversible posterior leukoencephalo-
pathy. Early CJD is characterized by increased DWI signal in cortex or deep gray matter (particularly the caudate nucleus and anterior putamen) or both (Figure 3). Intermediate CJD is characterized on DWI by progression of unilateral/asymmetrical lesions to greater contralateral/symmetrical involvement and progression of caudate lesions to involve the putamen. Late or terminal CJD is characterized by prominent generalized atrophy and ventricular dilatation. As an added benefit, DWI is more tolerant of motion artifacts than other sequences. This is an advantage since these patients usually suffer from myoclonic jerks. CSF 14-3-3 protein assay is useful in diagnosis of CJD. It is adjuvant rather than absolute test for diagnosis. Brain biopsy remains the gold standard diagnostic test for CJD. It typically shows spongiform changes.

No effective treatment has been identified for CJD, which is universally fatal.8 Mean survival time of sCJD is only 6 months. Care for patients with prion disease is supportive. Aim is to control myoclonus and seizures, and to provide nursing care.

CJD must be distinguished from other dementias. Occasionally, Alzheimer disease and frontotemporal dementia are associated with myoclonus and a more rapidly progressive course than is typical and are therefore, mistaken for CJD.9 Prominent ataxia or parkinsonism when present, may suggest dementia with Lewy bodies, progressive supranuclear palsy, or multiple system atrophy. Other entities that have been mistaken for CJD include paraneoplastic syndromes, demyelinating disease, sarcoidosis and Hashimoto’s encephalopathy.10

In view of the varied and non-specific presenting features of the disease in its early phase, it is important for physicians from different specialties to be cognisant of clinical and investigative features of CJD and its differential diagnosis. Radiology now plays a significant role in the diagnosis of CJD. In appropriate clinical setting DWI findings allow to suggest the diagnosis of CJD early in the course of the disease.

REFERENCES