A FAMILIAL FORM OF CREUTZFELDT-JAKOB DISEASE (CJD) IS LINKED TO THE D178N/V129 PRION PROTEIN (PRP) MUTATION

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A familial form of Creutzfeldt-Jakob disease (CJD) is linked to the D178N/V129 prion protein (PrP) mutation. We describe a patient, who in 2006, at the age of 47, manifested difficulties in left leg movements, cerebellar syndrome and mioclonic jerks followed, in few months, by severe cognitive impairment. Polygraphic EEG showed non-specific slowing of background activity and FLAIR and diffusion-weighted magnetic resonance imaging scans was typical for CJD diagnosis. The clinical picture slowly progressed attaining to a state of akinetic mutism in about two years. In the last three years of the disease, repeated severe febrile episodes for pneumonia and pneumonia "ab ingestis" led the patient to repeated hospital admission. She died in February 2012 for respiratory failure, after six years of disease.

We have obtained serial neurophysiological (full PSG and actigraphy) and neurovegetative (Tilt test and ECG based HRV analysis) during the disease progression.

PSG revealed marked abnormalities in the sleep profile, with total absence of vertex waves, spindles and K- complex. Sleep appeared very unstable with frequent transitions between wakefulness and light sleep, fragmented by frequent arousals associated with limb movements (Limb Movement Index, LMI = 64).

The neurovegetative assessment revealed normal results, both at tilt-test and HRV analysis, despite the severe clinical picture (akinetic mutism).

The PSG recordings, showing a "non-wake-non-sleep" sub-wakefulness state is consistent with a severe thalamo-cortical circuitry dysregulation. We hypotetize that the long survival of the patient, despite the relevant CNS involvement and the episodes of severe infections, is related to the conserved neurovegetative control.