

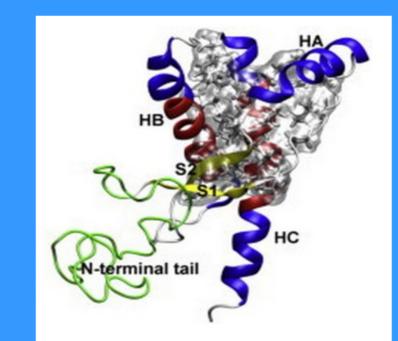
A case of genetic Creutzfeldt-Jakob disease presenting with insomnia



Giorgio G. Fumagalli, Paola Basilico, Marta Scarioni, Andrea Arighi, Anna Pietroboni, Daniela Galimberti, Nereo Bresolin, Elio Scarpini. IRCCS Ospedale Maggiore Policlinico di Milano, Università degli studi di Milano

Background

E200K is the most frequent cause of genetic Creutzfeldt-Jakob disease (CJD). Usually patients with this mutation present at the onset a rapidly progressive dementia, hallucinations or walking difficulties. Insomnia has been reported only later in the disease.



Methods

Fig. 1 (Mead S et al, 2006)

The authors characterized clinical, neuroradiological, neurophysiological and genetic features of the patient using MRI, EEG and prion protein (PrP) gene (PRNP) analysis

Family tree

History

The patient was born in a small village of Calabria of greek origin. In the same village other people, not directly related to the patient developed a rapid worsening dementia that quickly led to death.

In his family only the maternal grandmother died of dementia at 74 years old after 3 years history while his mother died of an ovarian tumor at 73 years of age.

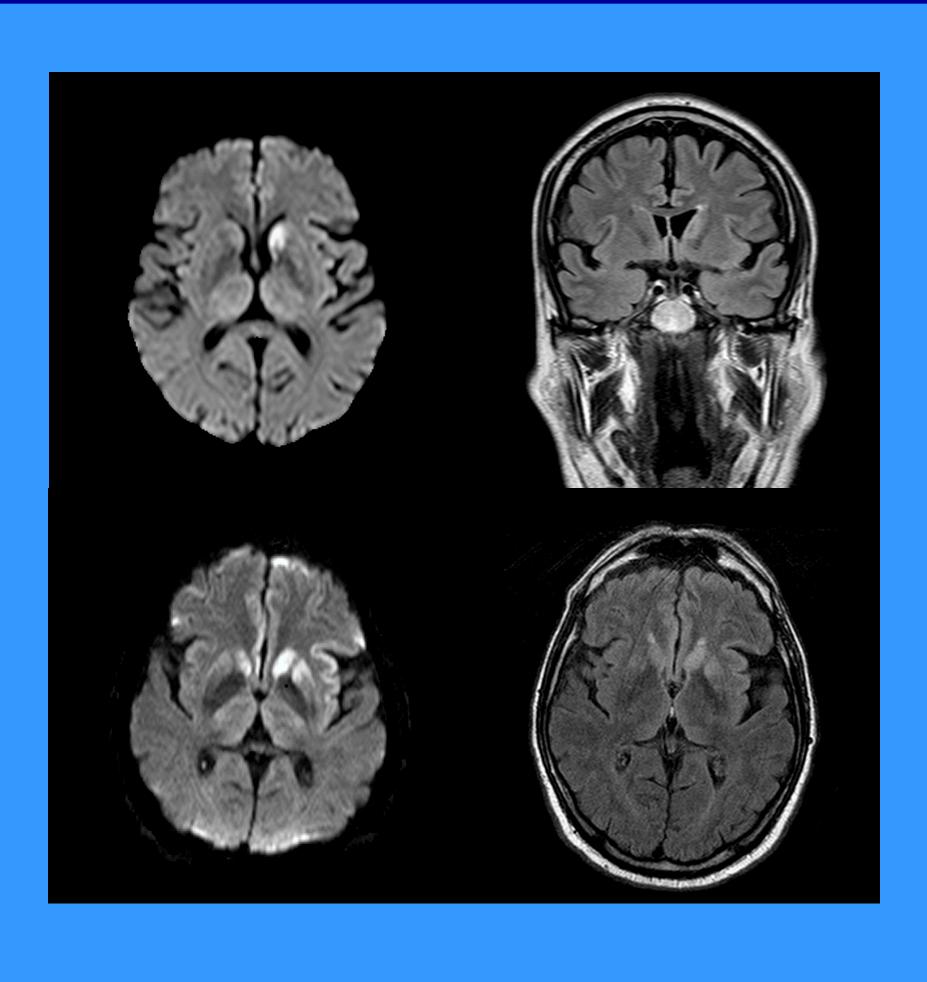
He had a history of hypertension and suffered of myocardial infarction at the age of 52. As a risk factor for prion disease relatives reported that he had eaten goat's brain multiple times.

Clinical picture at onset and development

The patient presented a drug resistant insomnia and this was the only symptom for one month. He later developed tremor at the beginning in right hand that rapidly spread to all four limbs. Then dysatria and gait disturbance. When he was referred to our hospital he had dysatria, disfagia, dysmetria, bilateral tremor and tone was increased bilaterally, more on the right limbs. Riflexes were brisk, with clonus in the right ankle. He had short term memory loss and was anomic. The following day he became bedridden, developed vertical gaze palsy, and the speech was unintellegible. He continued not to sleep and became agitated. In the third day presented negative myoclonus in the right arm, and VII nerve palsy. The forth day was drowsy and in the afternoon was no longer responsive with periodic breathing. The sixth day presented multiple tonicoclonic seizure and become febrile. He died eight days after the admission and three months after the first symptom onset.

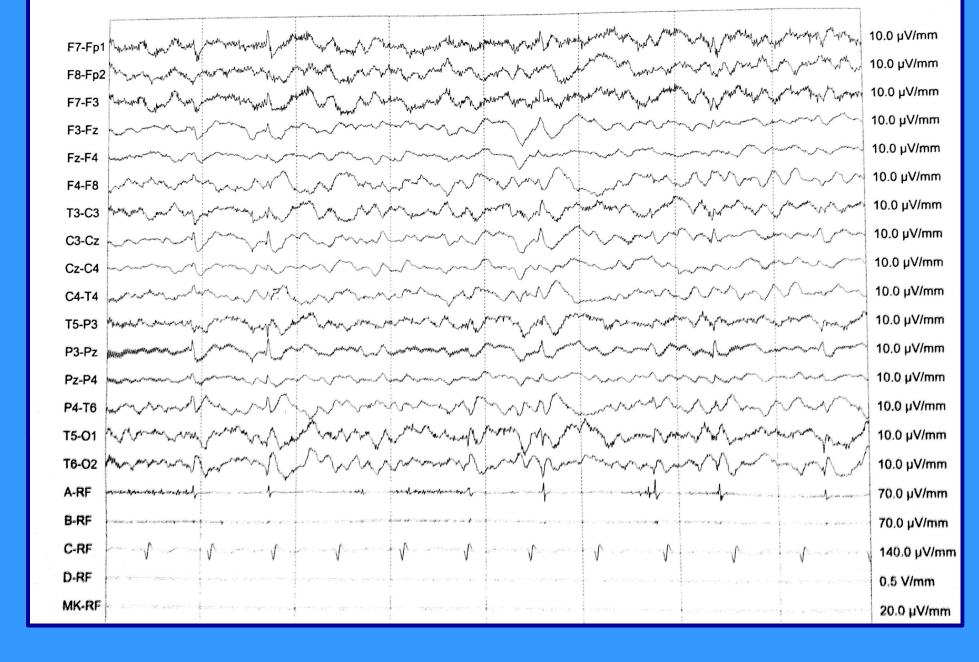
MRI

Brain MRI (DWI and Flair) one and three months after onset showing abnormalities in basal ganglia and talami bilaterally and cortical ribboning.



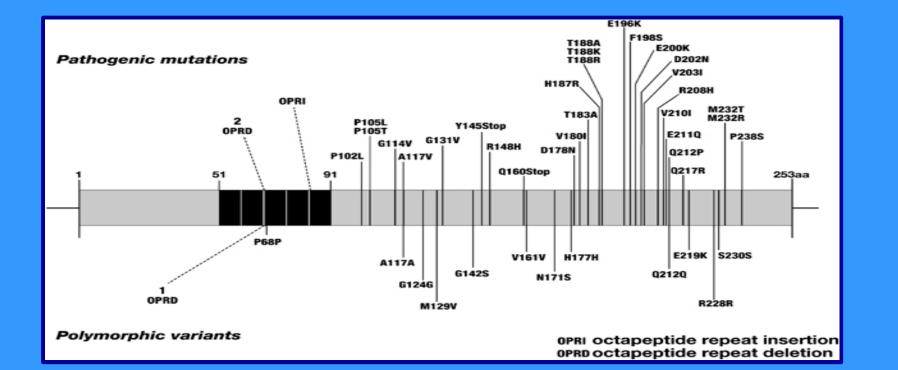
EEG

EEG showed sharp waves and periodism bilaterally with left prevalence.



Genetic

Genetic testing demonstrated a E200K mutation with Methionine-Methionine at codon 129 of PRNP.



Conclusions

Insomnia may be the first and only symptom at onset. This report confirms the heterogeneity of symptoms at presentation in genetic CJD

References

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