Creutzfeldt-Jakob disease: typical imaging findings

Ricardo Felix-Morais,[1] Luisa Costa Andrade,[1] Olinda Rebelo[2]

[1]Department Medical Imaging, Coimbra University and Hospital Centre, Coimbra, Portugal
[2]Department of Neurology, Coimbra University and Hospital Centre, Coimbra, Portugal

Correspondence to
Dr Ricardo Felix-Morais, ricardofelixmorais@gmail.com

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DESCRIPTION

A 63-year-old woman, previously healthy, presented with a 2-month history of progressive memory impairment, disorientation and behavioural changes. Examination revealed dysphasia, cerebellar ataxia and hypertonia of her upper limbs. Stimulus-sensitive myoclonus was also noted. Head CT was normal. MRI study was performed and revealed symmetric bilateral hyperintensities in putamen and caudate nucleus on T2-weighted (figure 1) images and fluid-attenuation inversion recovery (figure 2). On diffusion-weighted imaging, striatum and cortical frontotemporal gyriform hyperintense areas (cortical ribboning) were noted (figure 3), with restricted diffusion on apparent diffusion coefficient maps.

The rapidly progressive dementia associated with the typical imaging findings, was strongly suggestive of Creutzfeldt-Jakob disease (CJD).[1]

The disease progressed relentlessly, with rapid cognitive and functional impairment towards akinetic mutism. The patient died 3 months after her admission. Postmortem brain autopsy revealed the presence of abnormal protease-resistant prion protein (figure 4), confirming the diagnosis.

CJD, is a rare neurodegenerative disease, with an incidence of 1:1 000 000/year worldwide.[2] Sporadic CJD is caused by prion, an abnormal isoform of a normal host-encoded protein, leading to a spongiform encephalopathy.

The sporadic form is the most common (90%), but hereditary (10%) and acquired forms also exist (<1%).[2] Typically, symptom onset occurs at sixth to seventh decade, with rapidly progressive dementia associated with myoclonic jerks and a variable constellation of pyramidal, extrapyramidal and cerebellar signs.[3] Rapid progression towards

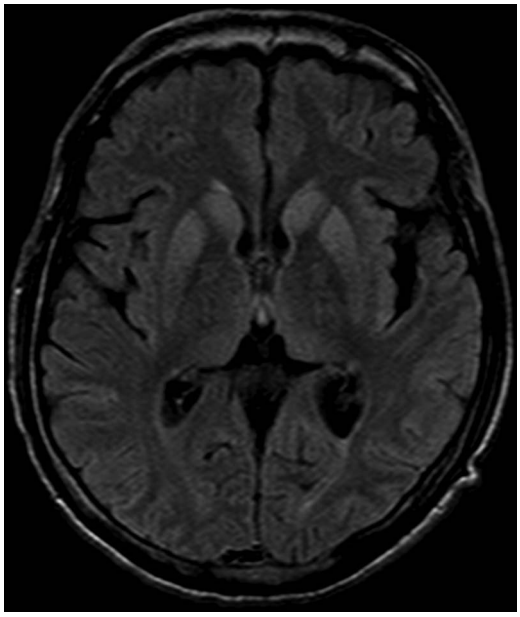

Figure 2 Axial fluid-attenuation inversion recovery: symmetric bilateral hyperintensity in the caudate and putamen.

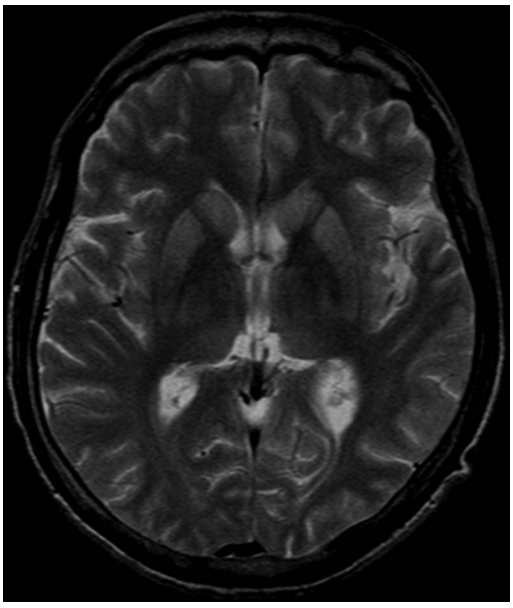

Figure 1 Axial T2-weigthed images: bilateral hyperintense signal in the caudate and putamen.

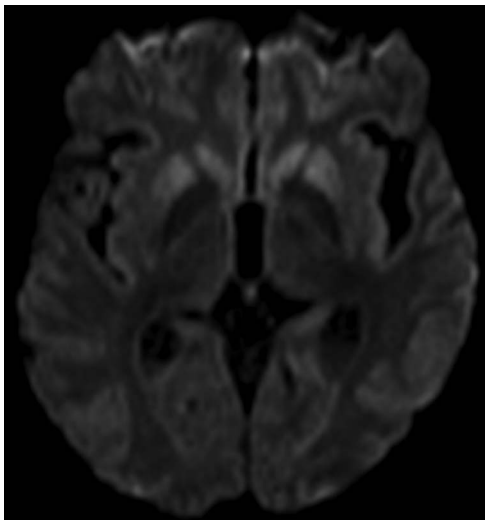

Figure 3 Diffusion-weighted imaging: diffusion restriction in the caudate nuclei and putamen. There is also typical hyperintensity in frontal and temporal lobe ('cortical ribbons').

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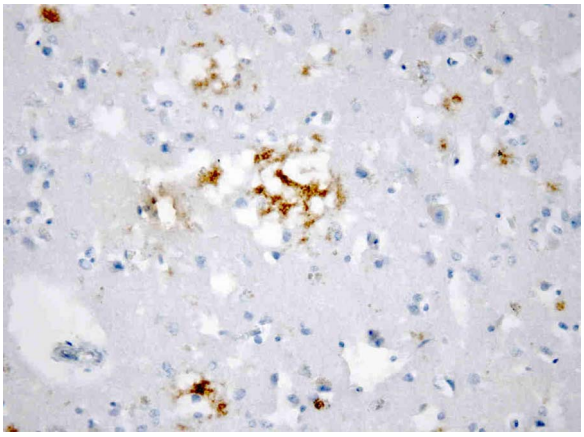

Figure 4 Immunostaining of protease-resistant prion protein.

akinetic mutism is the rule. Death usually ensues within months of clinical onset, with less than 10% rate survival within the first year.[2] No effective treatment exists.

Contributors RF-M and LCA wrote the final version of the article. OR realised the biopsy. RF-M, LCA and OR contributed with significant revision to the article. All authors read and approved the final manuscript.

Learning points

- A rapidly progressive dementia associated with myoclonic jerks, pyramidal, extrapyramidal or cerebellar signs, is suggestive of Creutzfeldt-Jakob disease.
- Best imaging tool: MRI with fluid-attenuation inversion recovery (FLAIR) and diffusion-weighted imaging (DWI).
- Best imaging clues
  - T2-weighted and FLAIR: bilateral hyperintense signal in basal ganglia.
  - DWI: hyperintensity in basal ganglia and cortex.

Competing interests None.

Patient consent Obtained.

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