

44th ANNUAL DIAGNOSTIC SLIDE SESSION 2003
CASE 2003-4

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Clinical history :

This 67 year old woman presented a ten-year history of transient episodes of hypersomnolence, dysarthria, ataxia, psychomotor slowing and documented hypothermia (32°C). These episodes were self-resolving. In addition, this patient had long standing constipation and severe sleep apnea. A complete metabolic work-up including analyses of cerebrospinal fluid was negative. Magnetic resonance imaging revealed marked atrophy of the brainstem. An EEG revealed the presence of frontal epileptic spikes; however the patient never had any clinical manifestations of epilepsy. Frontal deficits were evident on neuropsychological testing.

Several first-degree relatives of the patients were similarly affected. All shared with the patient a particular morphotype with marked kyphosis, arched palate and low hairline. The syndrome gradually progressed with dysphagia and dysarthria becoming increasingly severe. As well, cerebellar and pyramidal signs became more prominent. The patient ultimately died following complications of myocardial infarction at age 67. The brain weighed 1380 grams.

Material submitted :

- Kodachrome of transverse sections of the rostral medulla in the affected individual (left panel) compared to a normal patient (right panel) of similar age and brain weight (LHF/HE)
- H&E slide of either cerebellum or hypothalamus at the level of the optic chiasm.

Points of discussion :

1. Diagnosis
2. Pathogenesis