CASE 1997-2

Submitted by: Drs. Yves Robitaille, Stéphane Ledoux and Neil Cashman,
Ste-Justine and Montreal Neurological Hospitals,
University of Montreal, and McGill University,
3175, Côte Ste-Catherine Road,
Montreal, Qc., H3T 1C5, CANADA

Disease onset for this 19 y.o. right handed female, occurred during pregnancy as a brief episode of abnormal eye movements which, several months later, was followed by a resting tremor, first in the left hand, then in the right arm, both legs, head and tongue. Adequate dosage of L-Dopa brought no significant relief. Tremor was associated with marked fatigue, 40 pds weight loss and generalized joint pains. She also complained of episodic losses of vision in the left eye, which suggested a demyelinating disorder. The past medical history was characterized mostly by abnormal behavior, which led to the diagnoses of borderline personality disorder, anxiety, along with notions of alcohol and street drug abuse, although the latter could never be well documented. The family history could not be accurately expanded, but no specific notion of neurologic disease was recorded.

On examination, she was alert, oriented and there was no cognitive dysfunction. Insight and judgment were rated fair. Ocular movements showed slight limitation of upward gaze, and bilateral nystagmus on lateral gaze. Kayser-Fleisher rings were not seen. Peribuccal myoclonic movements and a tongue tremor were observed. A resting tremor was seen in all limbs, accompanied by a generalized increase of muscle tone and marked cogwheeling, but without significant loss of motor strength. She was described as "extremely" bradykinetic. DTR's were diffusely brisk, and the left plantar reflex was extensor. Besides a stooped posture, the gait was normal. Upper limb movements displayed decreased amplitude on walking. All lab data, which included an exhaustive work up to exclude a wide range of storage disorders, as well as CSF analyses, were within normal limits. MRI of the head was also normal, EMG studies revealed no pathology, but an EEG showed a generalized mild non specific slowing, suggestive of a sub-cortical dysfunction.

She was reassessed 18 months later. A thourough neuroendocrinologic work up was negative. She was diagnosed as an akinetic rigid disorder associated with behavioral abnormalities, consistent with a neurodegenerative disease of undetermined etiology. When last examined a few months prior to death, speech had become dysarthric, and spontaneous hand movements were consistent with myoclonic seizures, although, at times they appeared choreic, with occasionnal dystonic posturing. A few weeks before her final demise, she had become totally akinetic, diffusely hypotonic, hypotensive and mute. Unusual blood pressure fluctuations had been recorded throughout the course of the disease, however. She died suddenly during sleep at age 25 The immediate cause of death was ascribed to bilateral bronchopneumonia. A ubiquitinated section of frontal lobe was submitted.

Points for discussion: 1) Phenotypic diagnosis

2) Pathogenesis