bradykinesia, she was found to have porphyria. She had experienced conflicting symptoms concerning her porphyria,3 metabolisms. Most are intermittent and not always manifest, but her symptoms began to manifest as early as the age of 14. She had a medical history of dyskinesia which became remarkably sensitive to the size and frequency of the levodopa unit dose, especially in response to an increased exposure to dopamine receptor blocking agents and no autonomic symptoms. As her parkinsonism advanced she became increasingly depressed with hypochondriacic and paranoid delusions. She fulfilled the criteria for a severe depressive episode with psychotic symptoms (International Classification of Diseases (ICD) 10),3,4 and showed broad visual hallucinations and was noted to have a fluctuating level of consciousness.

Neurolological examination disclosed increased axial tone, impaired postural reflexes, and a shortened, stiff, and dystonic gait. She had a hypotonic saccadic eye movements, orofacial dyskinesias, and slow random alternating movements of her tongue. There was a resting tremor in all four limbs, associated with a symmetric akinetic rigid syndrome. On three hourly co-beneldopa (62.5 mg) she alternated between profound bradykinesias prelevodopa (motor subsection of the unified Parkinson's disease rating scale4 (UPDRS) = 44; Hoehn and Yahr stage 47), and a florid dyskinesia 30 minutes after her medication. The longest periods of her mood coincided with the periods of choreodyswastic movement and the absence of any evidence of pyramidal, cerebellar, or autonomic dysfunction.

The patient is one of 13 siblings. Two brothers and five other sisters have biochemical AI. One brother, a biochemical AI patient also fulfills UK Brain Bank criteria for Parkinson's disease (three hours after levodopa of 24 motor subsection UPDRS = 24). A maternal uncle was also diagnosed as having Parkinson's disease. Acanthoeytes were absent on a blood film. Urinary organic acid, amino acid, and copper studies were normal. No basal ganglia abnormalities were found on CT and MRI and autonomic function studies were within normal limits.

Benzhexol led to a modest improvement in her tremor. After this, pergolide was slowly introduced at a rate of 100 mg increments to 250 mg thrice daily. Her urinary pyrrolin concentration were monitored every 48 hours and remained unetectable. Her mobility and independence improved. There were asynchronous movements to a moderate improvement in her affect but the psychotic symptoms persisted. She responded well to six applications of electroconvulsive treat- ment (ECT) using propofol as the anesthetic agent. Over the subsequent eight months she did not develop any symptoms suggestive of acute porphyria and her urinary porphyrins remained within the normal range. Her psychotic depression relapsed within three months despite lopemipine maintenance but she again responded to further ECT.

This patient had idiopathic Parkinson's disease in addition to biochemical AI. The erratic and "brittle" response to levodopa treatment experienced by this patient may have been a result of porphyrin modulating the expression of the disease pathways. The therapeutic "window" for levodopa. Similarly, although porphyrin may have been responsible for or have influenced the expression of her affective disorder, the visual hallucinations and closing of consciousness raises the possibility of diffuse Lewy body disease, or an adverse effect of her anti-parkinsonian medication.

In some case reports, ECT has improved both bradykinesia and depression in patients with idiopathic Parkinson's disease, and it was effective in treating the patient's depression. Her parkinsonian syndrome improved while the pergolide dose was being increased without adverse consequences, providing some evidence to support the safety of pergolide in the treatment of porphyria.

Machado-Joseph disease is the most common autosomal dominant spinocerebellar degeneration. The pathogenic gene responsible for the disease, which was recently identified, contains a normal CAG expansion in the coding sequence of a novel protein, MJD1. The CAG expansion is inversely correlated with the age at onset, and is linked to other triplet diseases.

A case of Machado-Joseph disease presenting with spastic paraparesis

Machado-Joseph disease is the most common autosomal dominant spinocerebellar degeneration. The pathogenic gene responsible for the disease, which was recently identified, contains a normal CAG expansion in the coding sequence of a novel protein, MJD1. The CAG expansion is inversely correlated with the age at onset, and is linked to other triplet diseases. Four clinical subtypes of Machado-Joseph disease are well known. Type I patients show pronounced pyramidal signs and extrapyramidal signs such as dystonia, while Type II patients present with spastic paraparesis without dystonia or ataxia. A 38 year old woman was admitted to our hospital complaining of gait disturbance. She began to walk at the age of 1 year and 6 months. At the age of 7 years, she attended a school for mentally handicapped children. She walked on her toes, and sometimes she felt stiffness in her legs. At the age of 27 years, she became unable to go downstairs and walk without assistance.
Management of parkinsonism and psychotic depression in a case of acute intermittent porphyria.
P F Chinnery, N E Cartlidge, D J Burn, P G Cleland and I McKeith

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