CASE REPORT

Late onset dopa-responsive dystonia with tremor, gait freezing and behavioural disturbance and a normal dopamine transporter scan

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Abstract

A 79-year-old woman presented with dystonic posturing of the right leg while walking and an action tremor of her hands, both of which were levodopa responsive. She subsequently developed gait freezing. However, there was neither generalised bradykinesia nor rigidity. Structural imaging showed no significant changes, and a dopamine transporter scan was normal. She subsequently required rapidly escalating doses of levodopa in order to achieve symptom control, raising concerns over the possible development of a dopamine dysregulation syndrome. Issues raised included the difficulties of managing patients with a rare diagnosis and the role of dopaminergic medication with the potential for abuse.

Keywords: Parkinsonism, dopa-responsive dystonia, dopamine dysregulation syndrome, elderly, physician–patient relations

Case history

A 79-year-old woman presented with a 6-year history of her right foot turning inwards when walking and a tremor in the right hand. Subsequently, she froze in doorways and felt as though her feet were glued to the floor. There was a background of ‘jumpy legs’ for around 30 years. The condition was slowly progressive with considerable diurnal variation.

On examination, before starting dopaminergic medication, there was no facial amimia, bradykinesia or rigidity. On walking, her right foot turned increasingly inwards. There was a mild bilateral upper limb intention tremor, no other cerebellar features and normal eye movements. Initial investigations, including an MRI head, were unremarkable.

She was given a trial of levodopa for her dystonia. Her tremor, foot dystonia and gait freezing all markedly improved and she was able to remain active. However, she soon required increasing doses. Later, Pramipexole was added, again with rapid dose escalation. At this stage, she had a coarse tremor of her arms and legs on standing, and switched from sitting comfortably to almost bouncing with her whole body. She was clinically euthymic, though her mood dipped at times, but picked up with dopamine.

The clinical pattern raised concerns about the possibility of a psychological dopa-dependence syndrome, but this was rejected by the patient and her family, who perceived that she was being accused of substance abuse and threatened with the withdrawal of an effective medication. Not surprisingly, this strained the doctor–patient relationship.

The patient switched physicians and a dopamine transporter labelled scan (DaTSCAN) was arranged, which was normal. The patient agreed to trying lower pramipexole doses, though suffered dramatic deteriorations in physical and psychological symptoms. She reverted to her previous doses, with resolution of these symptoms.

With increasing awareness of adult-onset dopamine-responsive conditions with normal DaTSCANS, her diagnosis was revised to late-onset dopa-responsive dystonia with some features of dopamine dysregulation syndrome. Her symptoms are currently well controlled with moderately high doses of levodopa and pramipexole.

Discussion

This case raises issues inherent with a rare diagnosis, especially where there is a more common alternative explanation for the
Dopamine dysregulation syndrome affects ∼5% of Parkinsonian patients and is typified by addictive behaviour patterns, progressive tolerance to dopaminergic medication and significant withdrawal effects. It is thought to result from dopamine depletion in the mesolimbic system, reflecting the role of dopamine in mood and behavioural regulation [1,2].

A lot of the data on dopa-responsive dystonia come from paediatric reports from the Far East [3,4]. It results from abnormal dopamine synthesis in the striatum rather than neuronal loss. It typically presents with gait disturbance due to leg stiffness and an equinus gait (walking with the ankle in plantar flexion). There is marked diurnal variation and often minor features of Parkinsonism. The condition tends to deteriorate quite quickly over the first 15 years, and then stabilises. The stiffness and abnormal posturing affecting the legs can be so profound that it may resemble cerebral palsy. A dopa-responsive dystonia can be diagnosed with a dramatic response to low-dose dopaminergic therapy in the absence of other Parkinsonian features.

DaTSCANs are increasingly used to differentiate Parkinsonian disorders from other causes of tremor. Patients with late-onset dystonic tremors can look similar to Parkinson’s disease (PD), for example, with reduced arm swing asymmetrically when walking, slowness of movement (but no decrement suggesting true bradykinesia) and abnormal posturing [5]. However, in patients with dystonic disorders the DaTSCANs are normal, but are invariably abnormal in idiopathic PD [6].

The diagnostic and management difficulties in this case highlight the challenges of medicine and how new investigations can identify hitherto unrecognised conditions.

Key points

- Dopamine dysregulation syndromes affect ∼5% of PD patients.
- Rare diagnoses present a difficult management problem, especially where recognised disorders exist that could explain symptoms.
- This can lead to difficulties in the doctor–patient relationship.
- Technological developments can lead to the recognition of ‘new’ diseases.

Conflicts of interest

No conflicts of interest.

References


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