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Case Report

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Juvenile Parkinsonism – A Diagnostic Dilemma

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Abstract

Parkinson's disease (PD) is typically characterized by resting tremor, rigidity and bradykinesia with onset after 50 years of age. However sometimes, especially Juvenile Parkinsonism, may present with atypical features which make the diagnosis difficult and intriguing. Juvenile Parkinsonism i.e. onset of PD before 20 years of age, can present predominantly with dystonia and pain only without being associated with resting tremor. Here we report a case of Juvenile Parkinsonism in which diagnosis could be made only after several years of follow up and multiple consultations because of atypical features, and hence the diagnostic dilemma faced.

Keywords

Juvenile Parkinsonism, Movement disorder, Early onset Parkinsonism

Introduction

Parkinson's disease (PD) usually starts in middle-aged individuals, with a prevalence of about two percent after 65 years of age [1], but four to twelve percent of PD cases start before 40 years of age, termed as early-onset PD (EOPD) [2]. Quinn et al. [3], further divided EOPD in two groups; onset before 20 years was called Juvenile Parkinsonism (JP) while those with onset between 20 and 40 years were termed young onset Parkinson's disease (YOPD). They also mentioned that almost all patients of JP have a positive family history of Parkinson's disease. PD usually presents with resting tremor, bradykinesia and rigidity involving upper limbs after 50 years of age. However, there are certain atypical features which can delay or make the diagnosis difficult; such as juvenile onset, dystonia or rigidity starting in lower limbs and absence of resting tremor. We attempt to report here a case of Juvenile PD and highlight the existence of atypical onset in such cases and thereby justifying the need for high degree of suspicion one should keep while managing these group of patients.

Case Report

The index patient was a young male with onset of abnormal movements at the age of 12 years. These were characterised by gradual onset jerky movements of left leg, both at rest and while walking but not during sleep. The jerks would lead to difficulty in walking at times. Subsequently, he started complaining of pain in left leg which progressed gradually from being intermittent to continuous. He was first seen by a physician who advised certain investigations like complete blood counts and X-ray imaging of affected limb. All the reports came out to be

normal and he was advised calcium and multivitamins tablets, but the symptoms did not subside. Patient's family consulted few other physicians as well but to no relief. Over the course of several months, he began experiencing generalised weakness and inability to move his limbs and body, which lead to the patient being bed-ridden most of the times. Patient would also experience intermittent but prolonged contraction of his affected leg. There was no diurnal variation or any improvement in the symptoms after a period of rest or sleep. Patient was then taken to an orthopedician who advised magnetic resonance imaging of the lumbo-sacral spine as well as brain. These findings were also within normal limits and after several visits the patient was referred to the psychiatry department with a possibility of a functional illness i.e. conversion disorder.

The patient was first seen by the psychiatry department at the age of 17 years, i.e. five years after the illness onset. At the initial visit in the department of psychiatry, patient was assessed and all investigations were seen. Apart from the above symptoms, patient complained of restlessness, but did not report any abnormality in thought and affect. Owing to the non-specific nature of the complaints, he was diagnosed as a case of somatisation disorder and treatment was started accordingly. When on follow ups patient did not show any improvement, he was admitted in the psychiatry ward for re-evaluation of the diagnosis. Patient denied substance abuse or any past history of fever with seizure or loss of consciousness. General physical examination was unremarkable; however, central nervous examination was marked by bradykinesia, lead-pipe rigidity and occasional jerks of left leg. Cranial nerve examination was normal and there were no tremors or choreiform movements and no signs of cerebellar involvement were present. Kayser-Fleischer rings were not observed on ocular examination. He did not have any organomegaly and other systemic examinations were within normal limits. Complete blood count and microscopy was normal. Serum copper and ceruloplasmin levels were within normal limits. Mental status examination was unremarkable. Based on the history, physical examination and investigations, a provisional diagnosis of Juvenile onset Parkinson's disease was made with a differential diagnosis of Dopa-responsive dystonia. Patient's family did not give consent for genetic analysis of the index patient. Hence, he was presumptively started on Levodopa-Carbidopa combination. It was started at a low dose and titrated upwards resulting in gradual improvement with noticeable changes in gait and activities of daily living. Patient was then referred to the Neurology department for further treatment where the diagnosis was concurred. After few months of follow up in neurology clinic, patient had developed dose-peak dyskinesias forcing the treating team to reduce levodopa and substituting with agonist Pramipexole and Amantadine. Patient was followed up for over a year after that and had shown sustained improvement.

Discussion

In cases of early onset PD, the list of differential diagnoses is large as many illnesses have similar initial presentations such as, Dopa-responsive dystonia (DRD), Wilson's disease, Parkinson-plus syndromes, juvenile form of Huntington's disease, Neuroacanthocytosis, besides toxic and metabolic cause of Parkinsonism [4]. Wilson's disease was ruled out because of absence of Kayser-Fleischer rings on ocular examination and normal levels of serum copper and ceruloplasmin. This illness was distinguished from the Parkinson-plus syndromes like progressive supranuclear palsy and multiple system atrophy by the lack of loss of upward gaze or balance problems and absence of marked autonomic dysfunction, respectively. We could rule out most of the differentials in this case by the slow disease progression, normal blood investigations and neuroimaging studies as well as good response to levodopa therapy. Psychogenic movement disorders which are usually characterised by acute onset, variable intensity and frequency of movements reported and high suggestibility, could also be ruled out on the basis of history and clinical examination.

An important difference between DRD and EOPD is the presence, in DRD, of diurnal fluctuation (aggravation of symptoms toward the evening and alleviation of symptoms in the morning after sleep), a dramatic and sustained response (complete or near-complete responsiveness of symptoms) to relatively low doses of orally administered Levodopa and absence of motor adverse effects of long-term Levodopa therapy (wearing-off and on-off phenomena and Dopa-induced dyskinesias) under optimal doses of Levodopa [5].

The diagnosis of PD in this patient was difficult to make because of the several atypical features at initial presentation; such as younger age at onset, jerky movement and pain as the first clinical manifestation, absence of resting tremor, and initial involvement of the lower limbs. As mentioned earlier, 4 to 12% of PD cases start before 40 years of age as seen in early-onset PD [1].

Whereas most late onset PD present with tremors, bradykinesia and postural instability; early onset PD presents with rigidity [6]. In a prospective study done on 30 consecutive patients with EOPD at the National Institute of Mental Health and Neurosciences (NIMHANS), Bangalore; researchers found that as compared to young onset PD (YOPD), Juvenile Parkinsonism (JP) patients presented more with dystonia than resting tremors [7].

Index patient presented with initial symptoms in the lower extremities, a quite rare atypical feature in PD. Most often, PD starts in the upper limbs [8], but in about 20% of cases it can begin in the legs [9].

This case posed a diagnostic dilemma to the treating team because of these atypical features and after thorough history taking, investigations and examination, a diagnosis of Juvenile Parkinsonism was considered. The response to Levodopa clinically confirmed our diagnosis and brought relief to the patient, but not before the patient waiting for so many years and consulting several clinicians. Diagnosing and managing the index case was particularly challenging for him and his family both psychologically and socially. The response to the treatment came as a welcome respite to the patient and the family members. Parkinson's disease is a debilitating illness and awareness of these atypical features of early onset PD will certainly ensure reduction of physical and psychological morbidity in theses set of patients.

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