A professional sportsperson with subtle motor symptoms and signs: early-onset Parkinson's disease

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A 41-year-old professional hockey referee was referred to our clinic with an 8-month history of walking difficulties; previously he had consulted several orthopaedic surgeons and had undergone physical therapy—with limited benefit—for a presumed knee injury. The patient had no notable medical history, but reported two maternal aunts with Parkinson's disease which had presented at over the age of 65 years; his parents and sister were healthy. On examination he appeared generally fit and well; however, a neurological examination found that he had hypomimia, mild rigidity, and bradykinesia on motor tasks in both legs, but predominantely in the right (video). The patient's gait showed dystonic posturing of the right foot with involuntary plantar flexion and inversion, and asymmetrically reduced arm swing; his walking difficulties improved by running and walking backwards (figure; video). Additionally, despite having difficulties walking, he reported no problems while riding his bicycle (video).

Laboratory investigations showed no atypical findings, imaging likewise. Molecular genetic analysis showed compound heterozygous mutations of the parkin (*PRKN*) gene, confirming a diagnosis of early-onset Parkinson's disease. A levodopa and carbidopa combination, which was titrated up to 100 mg three times a day, provided sustained clinical benefit. At follow-up 6 months later, the patient reported feeling better, without fluctuations of motor function; the bradykinesia had improved and there was less dystonic posturing when walking. He had resumed his professional activities.

PRKN mutations are the commonest type of autosomal recessive Parkinson's disease, accounting for between 14 and 25% of early-onset cases. A delay between symptom onset and diagnosis is often observed because dystonia and parkinsonism may be unrecognised—especially in younger patients. Important clinical clues may be overlooked; for example, people with Parkinson's disease may find running or riding a bicycle easier than walking—as in our case—despite clear motor impairment, which is suggestive of dystonia.

The combination of parkinsonism with lower-limb dystonia in a young patient should alert clinicians to the possibility of Parkinson's disease with a genetic aetiology. Early-onset Parkinson's disease, caused by *PRKN* mutations, is a possibility, especially if hyper-reflexia, diurnal fluctuations, and levodopa-induced lower-limb dyskinesia are present.

Contributors
We were both involved in the patient's clinical care and management. We both acquired, analysed, and interpreted the data; and drafted the manuscript. RA provided supervision and a critical revision of the paper. Written consent for publication was obtained from the patient.

Declaration of interests
We declare no competing interests.

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Figure: **Searching for the subtle sign of dystonia in Parkinson's disease**
Photograph shows patient walking with dystonic posturing of the right foot with involuntary plantar flexion and inversion (curved arrow), and asymmetrically reduced arm swing.

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