SCA2 is an inherited, autosomal dominant neurodegenerative disease caused by an expanded trinucleotide repeat (>35 CAG) in the ATXN2 gene on chromosome 12q24.1. SCA2 typically manifests with progressive gait ataxia and dysarthria—similar to other spinocerebellar ataxias—but additional findings may include ophthalmoplegia, action tremor, dystonia, pyramidal tract signs, fasciculations, peripheral neuropathy, autonomic failure, cognitive dysfunction and parkinsonism.1

CASE REPORT

This 55-year-old Chinese woman presented to our clinic at age 46 with a 2-year history of progressive instability and impaired manual dexterity. Her initial physical examination revealed broad-based gait, mild dysmetria and facial fasciculations, later complicated by parkinsonian rigidity and bradykinesia. Her family history was notable for similar symptoms—predominantly ataxia—in 3 of her 5 siblings. MRI showed brainstem and cerebellar atrophy (Figure 1), and genetic testing confirmed spinocerebellar ataxia type 2 (SCA2) with 39 CAG repeats.

The patient was initiated on carbidopa-levodopa, which meaningfully lessened her parkinsonism; however, within three years she began to demonstrate wearing-off phenomena. After 6-7 years of treatment, she developed severe motor and nonmotor fluctuations, manifesting as peak-dose levodopa-induced dyskinesias (LID) with hypomania in the “on” state (Figure 2), alternating with off-state depression.

Lower doses of levodopa provided in closer intervals produced some improvement in her LID but resulted in intolerable “off” symptoms. Accordingly, the patient elected to return to higher doses despite the LID. Deep brain stimulation has not been feasible because of cognitive deficits.

References

9. a similar strategy may be warranted in patients with SCA2.