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INTERACTN CASE

The case of a 42-year-old man with progressive gait instability, dysarthria, and dysphagia

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Summary of Case

A 42-year-old man with a history of HIV on antiretroviral treatment presented to a hospital in Lusaka, Zambia with a 1-year history of progressive gait instability, dysarthria, and dysphagia. He was in his usual state of health until a year ago when he gradually developed unsteadiness when walking which quickly progressed to being unable to walk after 3 months. In addition, he had progressive difficulty with dexterity and controlling his hands, unable to feed himself 2 months after symptom onset. He also developed significant slurred speech and swallowing difficulty over the first few months. Now 1 year after symptom onset, his family brings him to the hospital due to concern about the rapid decline. The most recent CD4 count was 218 cells/mm^3, and the most recent HIV viral load was unavailable. On a neurological examination, his mental status is normal. He has choppy pursuits, slowed saccades, and gaze-evoked nystagmus in all directions. He has severe mixed dysarthria with a significant scanning quality, causing most of speech to be unintelligible. The examination is most notable for severe ocular, truncal, and bilateral appendicular dysmetria. He is unable to sit, stand, or walk independently. He has full strength with symmetric reflexes. Cerebrospinal fluid analysis was unremarkable (normal white blood cells, protein, glucose, bacterial, and fungal cultures). JC virus testing was not available. Brain CT was notable for significant bilateral cerebellar atrophy with relative preservation of white matter and a focal left cerebellar peduncle hypodensity (Fig. 1).

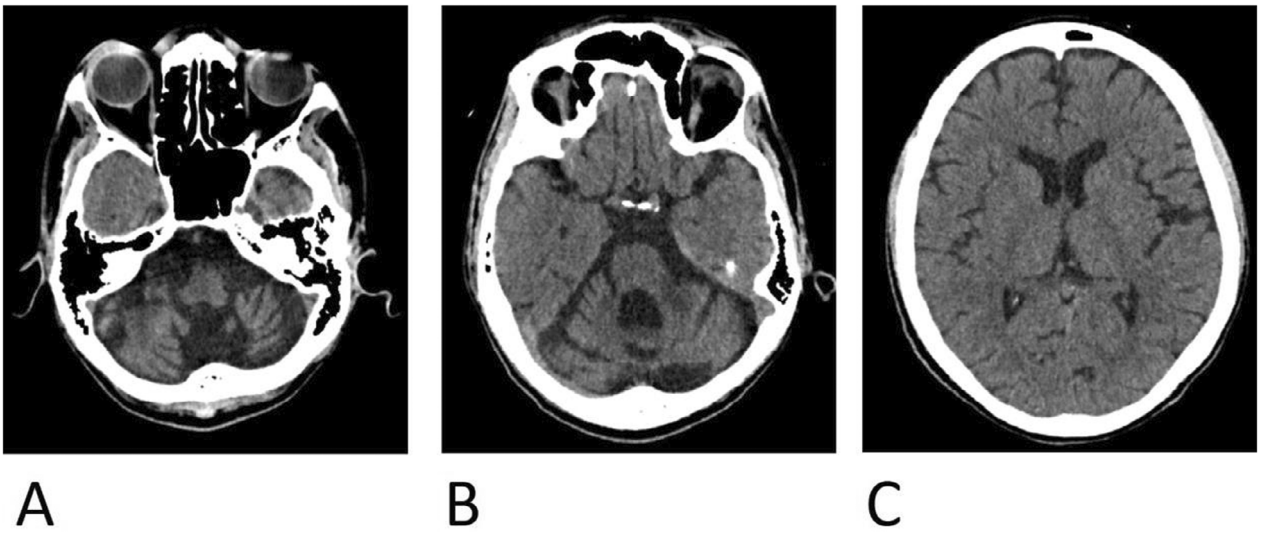

Figure 1. CT brain without contrast. (A and B) Profound generalized cerebellar atrophy. Also area of hypodensity in the left cerebellar peduncle. (C) No lesion or atrophy is seen in supratentorial regions of the cerebrum. Cerebellar atrophy notably out of proportion to supratentorial structures.

Diagnosis: Based on the available workup, the patient was diagnosed with presumed diagnosis of JC Virus Granule Cell Neuronopathy (JCV GCN) with possible left cerebellar peduncle progressive multifocal leukoencephalopathy (PML).

Take-Home Points

- In an immunocompromised patient presenting with clinical and radiologic signs of cerebellar atrophy with relative preservation of supratentorial areas, there should be a low threshold to consider JC virus granule cell neuronopathy (JCV GCN).
- Common opportunistic infections that can affect the cerebellum include tuberculosis, which can manifest as cerebellitis, and JC virus, which can cause progressive multifocal leukoencephalopathy (PML) affecting the cerebellar white matter and JCV GCN affecting the cerebellar granule cell layer.
- It is not uncommon that a definitive diagnosis is not possible in resource-limited settings. However, this case illustrates the execution of a systematic workup using available data from details of clinical presentation to available testing modalities to create a prioritized differential diagnosis that can guide empiric management.

