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Anti-Tr/DNER Cerebellar Ataxia after Immune-checkpoint inhibitors therapy in a patient with a SCA2 family history

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Introduction: Anti-Tr/DNER Antibody-Associated Cerebellar Ataxia is a classical paraneoplastic neurological syndrome (PNS) associated with Hodgkin lymphoma (HL).

Methods: Describe a case of a paraneoplastic cerebellar ataxia associated with the use of Immune-checkpoint Inhibitors (ICI) that was initially misdiagnosed as being hereditary.

Results: A 39-year-old woman presented to the ER complaining with a one-week history of unstable walking and diplopia.
Her past medical history revealed a HL diagnosis at 30 years of age successfully treated with first-line chemotherapy that relapsed 5 years later. She didn't respond to second-line chemotherapy and joined an experimental protocol with ICI, stopped 6 months before the onset of neurological symptoms. Her mother was affected by spinocerebellar ataxia type 2 (SCA2).
At the first evaluation she presented with minor neurologic signs; brain MRI was normal, so she was discharged with a diagnosis of suspected SCA2.
In the next few days she noted worsening of symptoms, therefore she came to our attention.
A new brain MRI showed increased volume of the cerebellar cortex. Laboratory analysis showed mild CSF pleocytosis and serum anti-Tr/DNER positivity.
Total body FDG-PET/CT didn't show HL relapses, so we treated her with steroids and IVIg without benefit. Now she is being treated with Rituximab and is slowly getting better. Genetic testing proved negative.

Conclusions: This case highlights the importance of excluding acquired and treatable causes of cerebellar ataxia before thinking of familial degenerative forms.
Furthermore, it could represent the first described case of neurologic Delayed immune-related adverse event (nDIRE) of ICI therapy.