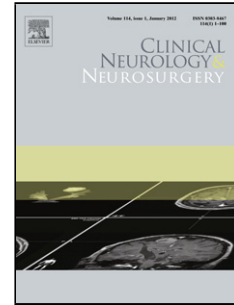


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Spastic paraparesis as the first manifestation of Machado-Joseph disease: a case report and review of the literature

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Highlights

- Machado-Joseph disease (MJD) is characterized by remarkable phenotypic heterogeneity.
- MJD manifesting as spastic paraparesis at the onset of disease (type V MJD) is rare.
- A MJD family with various phenotypes, including type V disease, in affected members is presented.
- A greater CAG expansion combined with an eastern Asian genetic background is associated with type V MJD.

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