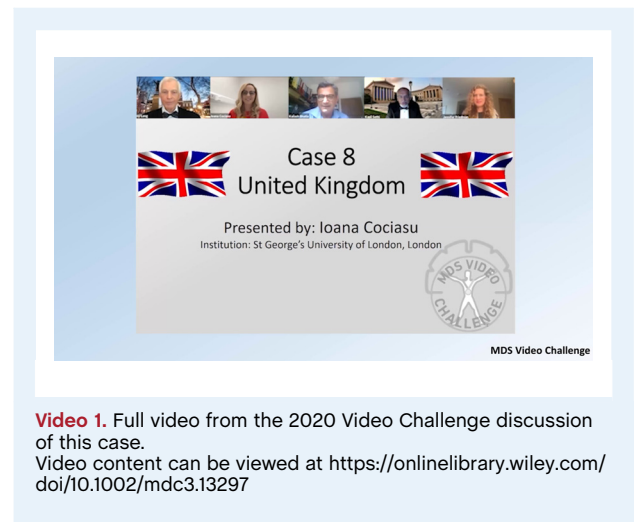


Commentary: Anderson-Fabry Disease: A Rare Cause of Levodopa-Responsive Early Onset Parkinsonism

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This case had young onset parkinsonism beginning at age 45 mainly affecting the right side with foot dystonia and limb pain as an early and prominent feature.¹ There was a family history of renal disease in one sister, stroke like episodes and dementia in another sister, and ischemic cardiac disease in the father. Single-photon emission computed tomography of the dopamine transporter showed bilateral reduced uptake and there was a good levodopa response and later development non-motor fluctuations. Although a number of parkinsonian conditions particularly genetic forms of young onset parkinsonism are in the differential, the patient also showed signs of renal (microalbuminuria) and cardiac (left ventricular hypertrophy) dysfunction which further narrowed the differential to conditions such as mitochondrial disease and neuronal inclusion body disease (though brain MRI did not show characteristic white matter changes). Given the severity of pain and renal involvement, Anderson-Fabry was considered as the most likely diagnosis. This x-linked lysosomal storage disease is caused by absent or minimal enzymatic activity of α -galactosidase and usually affects males in whom its fully penetrant but has been described rarely also to affect women.² It is important to recognize this condition given the availability of treatment with enzyme replacement therapy (agalsidase alfa). The list of genetic parkinsonian conditions is increasing.² Involvement of non-neurologic organ systems in patient or family members may offer clues to diagnosis. Anderson-Fabry disease specifically must be kept in mind in anyone with young onset parkinsonism with renal dysfunction and a pain (related to small fiber neuropathy).



Video 1. Full video from the 2020 Video Challenge discussion of this case. Video content can be viewed at <https://onlinelibrary.wiley.com/doi/10.1002/mdc3.13297>

Author Roles

(1) Research Project: A. Conception, B. Organization, C. Execution; (2) Statistical Analysis: A. Design, B. Execution, C. Review and Critique; (3) Manuscript Preparation: A. Writing of the first draft, B. Review and Critique.

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Keywords: Anderson-Fabry disease, early onset parkinsonism, lysosomal storage diseases, levodopa response.

Received 2 July 2021; accepted 6 July 2021.

Published online 00 Month 2021 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/mdc3.13297

Disclosures

Ethical Compliance Statement: The authors confirm that neither informed patient consent nor the approval of an institutional review board was necessary for this work. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this work is consistent with those guidelines.

Funding Sources and Conflicts of Interest: No funding was received for this work, and the authors have no conflicts to report.

Financial Disclosures for the Previous 12 Months: Ioana Cociasu, Chiara Sorbera, and Antonino Tuttolomondo have no disclosures to report. Francesca Morgante reports the following: Speaking honoraria from Abbvie, Medtronic, Bial, Merz, International Parkinson's disease and Movement Disorder Society; Advisory board fees from Merz and Bial; Consultancies fees from Boston scientific; Research support from Boston Scientific, Merz

and Global Kynetic; Royalties for the book "Disorders of Movement" from Springer; member of the editorial board of Movement Disorders, Movement Disorders Clinical Practice, European Journal of Neurology. Jennifer Friedman Participates in Clinical Trials with Biogen (Angelman's Syndrome) and Dr. Friedman's spouse is Founder and Principal of Friedman Bio-venture, which holds a variety of publicly traded and private biotechnology interests. ■

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