

Mystery Case: Lafora periodic acid–Schiff inclusion bodies

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A 16-year-old boy presented with a 5-year history of progressive cognitive decline and behavioral change followed by generalized tonic-clonic and myoclonic seizures refractory to many anticonvulsants (valproic acid, phenobarbital, clonazepam, and topiramate) and cerebellar ataxia a year later. The son of consanguineous parents, he had a family history of 2 cousins who both had epilepsy and died at the ages of 15 and 25 years. Laboratory screening tests, including lactic acid, fundus examination, brain neuroimaging, and CSF, were normal. Skin biopsy (figure) revealed periodic acid–Schiff-positive intracytoplasmic polyglucosan inclusion bodies in myoepithelial and sweat gland duct cells. These findings are typical of Lafora disease, a fatal autosomal recessive disorder caused by mutations in one of 2 known genes, both located at chromosome 6: *EPM2A*, which encodes the protein laforin, and *EPM2B*, which encodes the protein malin. Differential diagnosis must be made with other causes of progressive myoclonic epilepsies, most commonly Unverricht-Lundborg disease (Baltic myoclonus), myoclonus epilepsy and ragged-red fibers syndrome, neuronal ceroid lipofuscinosis, and type 1 sialidosis. There is no treatment, and therapy is mainly supportive and symptomatic.^{1,2}

AUTHOR CONTRIBUTIONS

All authors contributed equally to the study concept and design, acquisition of data, analysis and interpretation of data, drafting of the manuscript, critical revision of the manuscript for important intellectual content, and administrative, technical, and material support.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

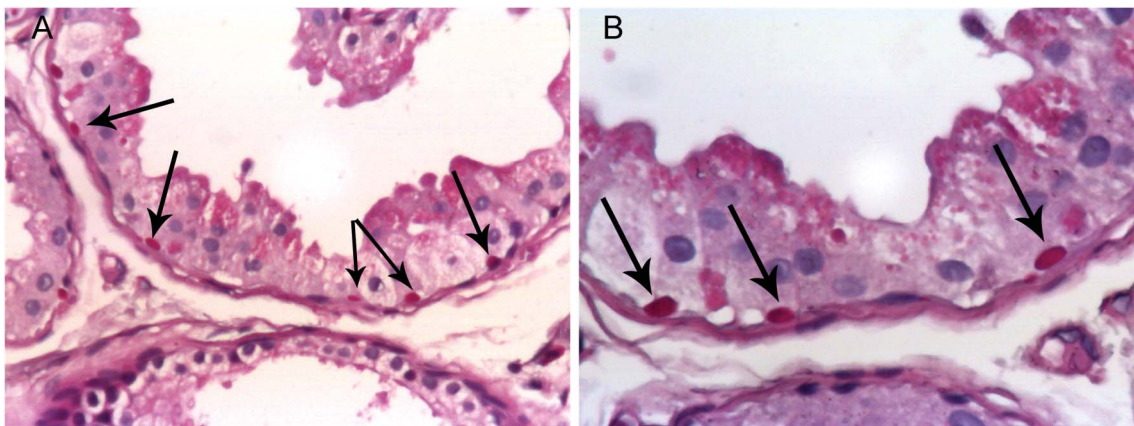
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MYSTERY CASE RESPONSES

The Mystery Case series was initiated by the *Neurology*[®] Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses were solicited through a group e-mail sent to the American Academy of

Figure Lafora bodies on skin biopsy



(A) Histopathologic examination of the skin biopsy taken from the axillary region reveals round-to-oval intracytoplasmic, periodic acid–Schiff-positive, diastase-resistant inclusions within the myoepithelial and acinar cells of sweat glands, corresponding to Lafora bodies (arrows). Periodic acid–Schiff stain, 200 \times . (B) Lafora bodies in a magnified view (arrows). Periodic acid–Schiff stain, 400 \times .

From the Neurology Service (I.d.A.F., E.T.d.R., H.A.C.d.R., M.P.P., L.A.P., T.C.V.) and Pathology Service (N.M.d.S.G.), University Hospital, Department of Internal Medicine, Faculty of Medicine, Federal University of Juiz de Fora (UFJF), Juiz de Fora, Minas Gerais, Brazil.

Neurology Consortium of Neurology Residents and Fellows and through social media.

All of the respondents correctly identified the Lafora bodies in the figure and concluded that the underlying diagnosis is Lafora disease. Eighty percentage of respondents accurately stated that this is an autosomal recessive disease associated with mutations in the *EPM2A* and *EPM2B* genes.

This case illustrates the value of a detailed family history in guiding the appropriate diagnostic tests for rare genetic disorders such as the progressive myoclonic epilepsies.

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