Retinitis pigmentosa in Lafora disease

Expanding findings of progressive myoclonic epilepsy

Figure Lafora disease and retinitis pigmentosa

(A) Funduscopy examination discloses retinitis pigmentosa. (B, C) Axillary skin biopsy discloses the pathognomonic periodic acid-Schiff–positive inclusion bodies, the so-called Lafora polyglucosan bodies, in the eccrine ducts of the sebaceous glands (arrow). (D) Sagittal T1-weighted brain MRI shows mild atrophy of the upper portion of cerebellar vermis.

A 21-year-old man presented with a 4-year history of seizures, visual hallucinations, cognitive decline, and gait impairment. Neurologic examination revealed myoclonic jerks, ataxia, and retinitis pigmentosa. Axillary skin biopsy showed Lafora bodies (figure). Lafora disease, the most common progressive myoclonic epilepsy with adolescent onset, is characterized by cognitive decline, visual hallucinations, myoclonus, generalized seizures, and pathognomonic inclusion bodies of polyglucosan found in cells of the skeletal muscle, skin, and brain.^{1,2} Retinitis pigmentosa is a hereditary pigmentary retinopathy commonly present in neurologic disorders such as mitochondrial diseases, abetalipoproteinemia, and Refsum disease³; however, it has never been described in Lafora disease.

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