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.
Retinitis pigmentosa in Lafora disease: Expanding findings of progressive myoclonic epilepsy
Wladimir Bocca Vieira de Rezende Pinto, Paulo Víctor Sgobbi de Souza, Jhonatan Rafael Siqueira Pinheiro, et al.

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