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Diagnosis of Lafora Body Disease by Axillary Skin Biopsy

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Abstract: Lafora body is a rarely seen and progressive disease which is characterized by mental decline, myoclonus and generalized epilepsy. Definitive diagnosis is made with biopsy showing typical spherical PAS positive inclusion bodies. In this article, we present a case who had myoclonus, generalize seizure and dementia and diagnosed with Lafora body disease. Diagnosis was confirmed by axillary skin biopsy. Na Valproat 15 mg/kg was started by orally for myoclonic and generalized epilepsy. Partial improvement in both myoclonic and generalized seizure frequencies was seen.

Key Words: Progressive myoclonic epilepsy, generalized epilepsy, mental decline

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