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Diagnosis of Lafora disease in axilla skin biopsy: Two case reports

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Introduction: Lafora disease is a fatal neurodegenerative condition characterized by presence of inordinate accumulation of Lafora bodies, which are insoluble glycogen-derived (polyglucosan) particles. Lafora disease are caused mostly by mutations in EPM2A and EPM2B. Diagnosis can be made by skin, liver, muscle and brain biopsy that reveals Lafora body deposition.

Case Report: Our first patient was a 47-year-old female who was previously healthy until she was 17-year-old when she experienced a seizure attack. Her seizures were refractory to anti-convulsive therapy and the disease resulted in worsening of upper and lower extremity movements. Subsequent progressive neurological deterioration lead to a decrease in ability to perform her daily activities. Family history revealed that her parents were second-degree relatives. Our second patient was a 19-year-old female who was previously healthy until she was 16-year-old when she exposed to a head trauma which after one and a half months give rise to the development of a myoclonic seizure attack. Light microscopic findings of axilla skin biopsy of the two patients revealed PAS-positive and diastase-resistant intracellular inclusion bodies located in the basal side of the apocrine acini. In the first patient, after monitoring for thirty years with treatment-refractory seizure, aksilla skin biopsy is taken which showed Lafora bodies. Second patient is under control with treatment for five months after diagnosis.

Conclusion: Diagnosis of Lafora disease is used to made with cerebral cortex biopsy which was quite an invasive procedure. But today, the preferred biopsy site is axilla skin that can be reached easily for biopsy and contains abundant apocrine sweat glands where Lafora bodies could be detected as PAS-positive inclusions. Electroencephalographic studies remain undoubtedly non-specific for the diagnosis of Lafora disease; however, detection of Lafora bodies which are not seen with any other progressive myoclonic epilepsy, is used as a pathognomonic feature for definite diagnosis.

Anahtar Kelimeler : Lafora disease, epilepsy, apocrine, axilla