

## 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

### Aksiller Cilt Biyopsisi İle Tanı Alan Lafora Body Hastalığı

**Özet:** Lafora body nadir görülen, ilerleyici, mental yıkım, myoklonus ve jeneralize epilepsi ile karakterize bir hastalıktır. Kesin tanısı biopsi materyalinde tipik PAS pozitif inklüzyon cisimciklerinin görülmesiyle konur. Biz bu makalede demans, generalize nöbet ve myoklonusla prezente, kesin tanısı aksiller cilt biyopsisi ile doğrulanan bir olgu sunmaktayız. Sodyum valproat oral yoldan 15 mg/kg jeneralize nöbetler ve myoklonus için başlandı. Jeneralize nöbet ve myoklonus sıklığında kısmi iyileşme elde edildi.

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**Anahtar Sözcükler:** İlerleyici myoklonik epilepsi, jeneralize epilepsi, mental yıkım

### Introduction

Lafora body (LB) is an autosomal recessive hereditary disease characterized by progressive dementia, myoclonus and generalize seizure (1-13). It was first described as a progressive myoclonus epilepsy by Lafora and Gluech in 1911 (8). Symptoms mainly begin in the first and second decade of life between 6 to 20 years (1,2,4,8). Initial clinic findings various, but a generalized seizure is the first symptom in the majority of cases. The diagnosis is confirmed by the demonstration of typical PAS positive spherical inclusion bodies in the brain and spinal cord, skin, liver and skeletal muscle on biopsies (1-6,8,10). For diagnosis, axillar skin biopsy is preferred being less invasive and gives lower false negative results (1,2,6,8,10). In this article, we present a case who had mental decline and a myoclonus-generalized seizure, with a positive family involvement.

### Case Report

A 16 years old, male patient was admitted to the hospital with myoclonus, generalized tonic-clonic seizure and mental decline. His past history revealed a tonic-clonic seizure three years earlier, followed by myoclonus involving the left arm and leg 6 months later, and finally, a progressive mental decline such as forgetfulness, personality deterioration, cognitive dysfunction and dissociated crying or smiling episodes in the last year. He had a positive family history; his two siblings had the same symptoms and both of them were lost after 5 and 7 years of the onset of the disease, respectively. On physical examination; his blood pressure, heart rate and systemic functions were all within normal limits. Neurologic evaluation revealed; partial cooperation and orientation, with occasional absurd behaviour. Cerebellar, pyramidal, extrapyramidal and peripheral nerve system findings were all normal. On laboratory investigation, routine blood, urine and biochemical tests and magnetic resonance (MRI)

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imaging were normal. Electroencephalography (EEG) evaluation showed diffuse and non localized multiple spikes and slow wave forms. His mini mental state was near debilitated levels. Biopsy of axillary sweat gland duct cells showed polyglucosan bodymatter which was characteristic of LB disease (Figure:1A-1B). Na valproat 15mgr/kg/day was started by orally. Both myoclonus and generalized seizure frequency were decreased. Additionally, cooperation and mental decline recovered slightly.

The etiology of LB disease is unknown and it affects both sexes equally (1,2,4,6,8). Although polyglucosans inclusions are characteristic of this disease, as of yet no enzymatic deficiency or abnormality in carbohydrate metabolism has been demonstrated [8]. In most of the cases, generalized seizures are the first symptoms of the disease. Mental decline starts usually later in the course of the disease, but it can rarely also be the initial finding (1,4,8). Kaufmann et al reported that epileptic seizures can be responsible for personality deterioration and

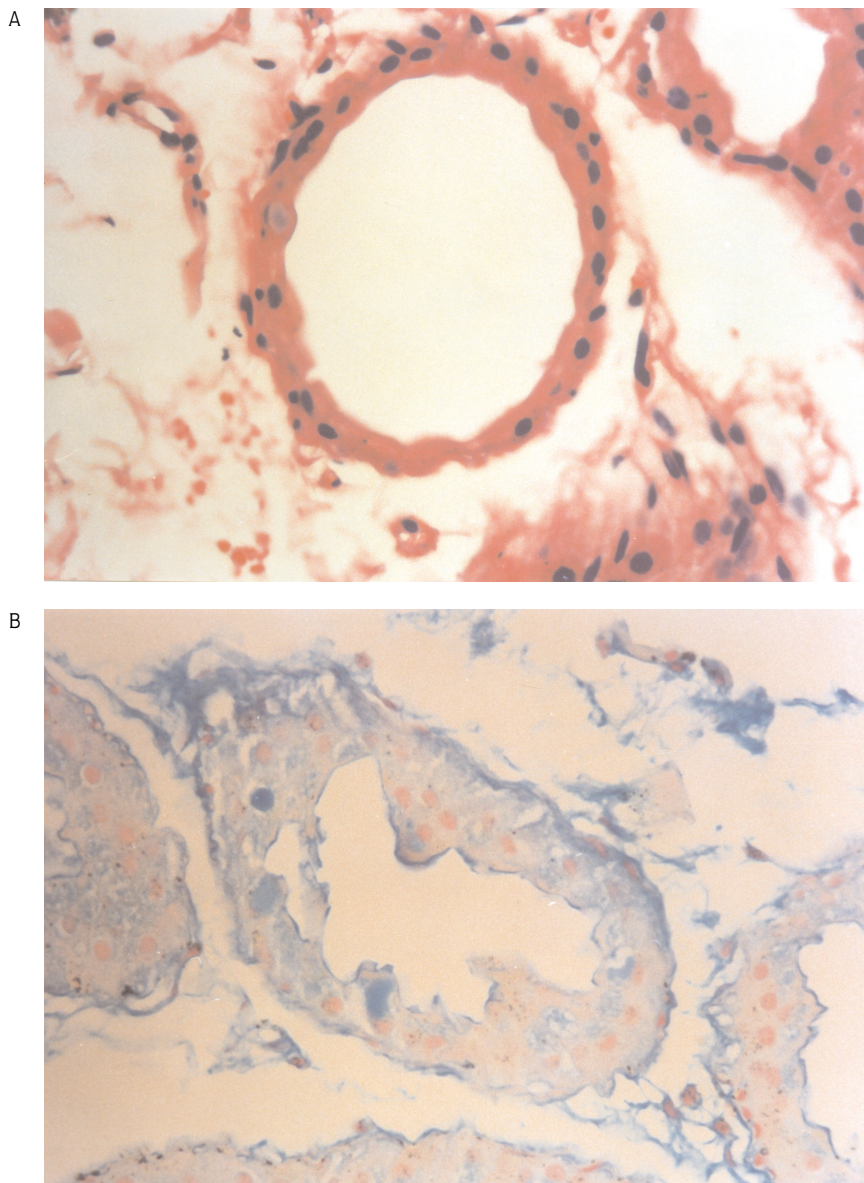


Figure 1. Axillar ductal biopsy with hemotocsilen eosinofil (A), polyglucosan spherical PAS-positive inclusion bodies in axillary sweat gland duct cells (B).

mental decline (4). Our case's clinical courses also supported this opinion that mental functions were partially improved after seizure frequency decreased. Visual ictal phenomena appear in half of the cases and are a relatively specific clinical clue to the diagnosis of disease (1,4,12,13), but this clinic feature was absent in our case. The characteristic EEG pattern consists of slow background recurrent epileptiform discharges including spikes, polyspikes, spike-wave and polyspike-wave complexes. Additionally, it has been shown that EEG remains almost unchanged with disease progression (1). In the presented case, the patient's EEG showed diffuse and nonlocalized polyspike and slow wave formation.

The diagnosis may be confirmed by the demonstration of typical spherical PAS-positive inclusion bodies in the brain and spinal cord, heart and liver, skeletal muscle and axillary sweat gland duct cells (1-4,8,10). Those inclusions are polyglucosan bodies which are not specific for LB disease. Similar changes can also be seen in certain conditions and diseases such as normal aging, type IV glycogen storage disease, arylsulfatase A pseudodeficiency, some instances of amyotrophic lateral sclerosis and upper or lower motor neuron disease. Myoclonus bodies are often seen in neuropil and neuronal perikarya in LB (4), but locate in axons and astrocytes in adult polyglucosan disease (8,10). Corpora amylacea

which is a normal feature of the aging brain, imitates Lafora bodies, but it is not found in neuronal structures (6).

In the differential diagnosis; subacute sclerosing panencephalitis (SSPE), progressive myoclonic ataxia (PMA), progressive encephalitis (GM2 gangliosidosis, Nieman Pick, Gaucher disease), juvenile myoclonic epilepsy, nonketotic hyperglycemia should be considered. Inheritance pattern, absence of burst-suppression on EEG and typical biopsy findings; presence of mental decline and tonic-clonic seizure; and normal blood glucose levels help differentiate SSPE and progressive encephalitis; PMA; juvenile myoclonic epilepsy; and nonketotic hyperglycemia, respectively.

Antiepileptic drugs, especially Na valproate are preferred for the treatment of both myoclonic and generalized seizures. We also used Na valproate in our case. Both myoclonic and generalized seizure frequencies diminished. The majority of cases die six years after the onset of symptoms which usually occur between 16-24 years of ages (1,2,4,6,8,11). In our case's siblings died at the ages of 15 and 17.

In conclusion, we can say that the aim of this article is to remind LB disease and importance of non invasive axillar skin biopsy.

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