

A Case Report of Alpers syndrome in a 6 year old girl presenting with Status Epilepticus

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Abstract

Introduction: Alpers syndrome is a mitochondrial disease caused by a mutation in the POLG-1 gene, while the symptoms are typically a triad of liver failure, epilepsy and progressive developmental regression. Status epilepticus is most often the first symptom.

The Case: A 6 year old girl with a background of epilepsy and mild developmental delay presented to the emergency department with refractory status epilepticus. The patient had suffered from tonic-clonic seizures from 4 years of age, for which she was being sufficiently treated with lamotrigine and clobazam. She started walking at 22 months (delay), and on two prior occasions was found to have muscle stiffness in her legs. A full blood count, coagulation profile and liver function tests were carried out, all of which were normal apart from a mild elevation in aspartate amino transferase levels. Despite a normal magnetic resonance imaging scan, the electroencephalogram showed slow delta waves with superimposed spikes. Although the patient's family history was unremarkable, next generation sequencing (NGS) testing was carried out which found both parents to be carriers for POLG mitochondrial cytopathy.

Outcomes: The diagnosis of Alpers syndrome in this patient was difficult, due to the lack of specificity of her past medical history.

Biography:

Yasmin Zolfaghari is currently in 4th year of medical school and have previously achieved a first class honours iBSc in Public Health. She published one systematic review in the past and also in the process of publishing 2 more. She is an aspiring paediatrician and hopes to invoke change with regards to research in paediatric rare diseases

Speaker Publications:

1. Status epilepticus in children with Alpers' disease caused by POLG1 mutations: EEG and MRI features, December 2008 *Epilepsia* 50(6):1596-607
2. Alpers-Huttenlocher syndrome: A review, *Pediatr Neurol.* 2013 Mar; 48(3): 167-178

[2nd Pathology and Infectious Disease Conference](#); Webinar- December 14, 2020.

Abstract Citation:

Yasmin Zolfaghari, A Case Report of Alpers syndrome in a 6 year old girl presenting with Status Epilepticus, Pathology Meet 2020, 2nd Pathology and Infectious Disease Conference; Webinar- December 14, 2020

<https://biopathology.pathologyconferences.com/2020>

