Insight Medical Publishing

Journal of HIV & Retro Virus

ISSN: 24719676

2020

Sp.Iss.111

# A case report of Alpers syndrome in a 6 year old girl presenting with status Epilepticus

Yasmin Zolfaghar

Barts and the London School of Medicine and Dentistry

***Abstract:***

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:***

I am in my 4th year of medical school and have previously acheived a first flass honours iBSc in Public Health. I have published one systematic review in the past and am in the process of publishing 2 more. I am an aspiring paediatrician and hope 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 2008Epilepsia 50(6):1596-607

2. Alpers-Huttenlocher syndrome: A review, Pediatr Neurol. 2013 Mar; 48(3): 167–178

3rd Annual Conference on Bacterial, Viral and Infectious Diseases; Dubai, UAE - June 18-19, 2020.

**Abstract Citation:**

Yasmin Zolfaghar, A Case Report of Alpers syndrome in a 6 year old girl presenting with Status Epilepticus, Bacterial Diseases 2020, 3rd Annual Congress on Bacterial, Viral and Infectious Diseases; Dubai, UAE, June 18 -19, 2020.

(https://bacterialdiseases.infectiousconferences.com/2020