THE EFFECT OF CLADRIBINE TABLETS ON DELAYING MOTOR NEURON DISEASE WITH MALIGNANCY: CEREBELLAR OEDEMA IN FULMINANT ADULT LEIGH SYNDROME

Introduction

While some regard an association between motor neuron disease (MND) and malignancy as coincidental, others report a case of adult Leigh syndrome resulting in fulminant cerebral oedema.

Case

A 19-year-old female presented with a five-week history of hyperventilation, generalised weakness, dysarthria and bilateral ptosis. Brain Magnetic resonance imaging (MRI) findings and the presence of a mitochondrial mutation (NC_012920.1 (MT-ATP6):m.9176T>C) in blood and urine with approximately 97% heteroplasm, confirmed a diagnosis of Leigh syndrome.

Two-days after a normal lumbar puncture, opening pressure 8cm water, her conscious level rapidly declined. CT revealed marked cerebellar oedema with brainstem compression. Despite immediate decompression, she did not recover consciousness and died six-weeks after symptom onset.

Conclusion

Adult Leigh syndrome is a progressive untreatable inherited mitochondrial disorder typically of infants and children. Adult cases are rare and described mostly in single case reports. There is marked phenotypic and genotypic variability. Over 83% of Leigh’s syndrome is identified by the age of 2 years, however, there have been cases reported in patients up to 74 years old. There are over 60 mutations described in Leigh syndrome, which are identified in only half of reported cases. Classic MRI changes include bilateral symmetric T2 hyper-intensities in the basal ganglia and brainstem. To our knowledge, this is the first reported case resulting in fulminant cerebellar oedema. A challenge of diagnosis remains the marked heterogeneity in presenting symptoms including cognitive decline, behavioural change and ophthalmoplegias. Typically, this syndrome has been confirmed by histopathology at autopsy. Advances in genetics and imaging have allowed earlier accurate diagnosis, potentially paving the way for improved therapeutics.