THE INTENSIVE CARE JOURNEY OF A CHILD WITH UNIQUE CALCIUM CHANNEL MUTATION WITH SARS-CoV-2

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Introduction

We describe the first reported case of a child with early onset Developmental and Epileptic Encephalopathy (DEE), associated with a de-novo nonsense mutation in CACNA1G.

We outline his acute presentation, explore the paediatric intensive care unit (PICU) journey and reflect on the impact of the COVID-19 pandemic.

Case summary:

AB was a term infant born by emergency caesarean section who developed daily myoclonic and generalised clonic seizures at 3 months of age.

Whole genome sequencing on DNA from AB and both parents showed a de novo nonsense mutation.

At 35 months AB presented with respiratory distress and seizures requiring invasive ventilation. A blind broncho-alveolar lavage (B-BAL) grew Acinetobacter baumannii, Moraxella catarrhalis and Escherichia coli. After multiple unsuccessful extubations, a decision was made to proceed with a tracheostomy. At this time the COVID-19 pandemic developed and AB became pyrexic with a rise in inflammatory markers. Viral PCR on B-BAL identified “severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2)”. AB steadily improved with supportive care.

After 121 days of ventilation a final attempt at extubation was made due to collaborative decision making between parents and clinicians being mindful of risk/benefits but being cogniscent of delayed ENT operating in the setting of COVID-19. AB successfully extubated.

During his stay in PICU, AB had a fragility fracture of his left femur managed with leg cast and medical therapy.

Conclusion:

The case describes the presentation and clinical journey of a child with a previously undocumented condition.

Decision making regarding direction of care was challenging due to the unique nature of the case.

AB acquired COVID-19 infection whilst being in PICU despite infection control precautions highlighting the virulence of the disease.