Acetazolamide for severe early onset epileptic encephalopathy due to mutations in the *KCNA2* gene

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Background: *De novo* loss or gain of function mutations in *KCNA2* gene have been described in individuals with epileptic encephalopathy, ataxia or intellectual disability. Seizures are usually refractory to antiepileptic medications.

Aim: Access the effect of acetazolamide on patients with early onset epileptic encephalopathy caused by *KCNA2* gene mutation both clinically and electro-physiologically.

Methods: We report a cohort of 11 patients with severe early onset epileptic encephalopathy carrying *KCNA2* mutations. All had refractory seizures resistant to multiple antiepileptic drugs, significant developmental delay and slowing of EEG background. We started them on acetazolamide after antiepileptic medications failed. Pre and post therapy seizures burden and electroencephalography (EEG) studies were evaluated.

Results: 9 of the 11 children (81%) showed a significant improvement both clinically and electro-physiologically and Acetazolamide is a potentially effective therapy in patients with early onset epileptic encephalopathy carrying *KCNA2* mutations.

Biography

Khalid Hundallah is a Consultant Pediatric Neurologist. He is the head of Pediatric Neurology, Prince Sultan Military Medical City. He is an Assistant Professor, Colleague of Medicine, Al-Imam `Mohd Ibn Saud University. He has completed his fellowship in McMaster University. He has published more than 18 papers in reputed journals and has been serving as an Associate Editor in the Neuroscience Journal (KSA).

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