SPTAN1 related epileptic encephalopathy- a case study

Hazem Eltoukhy[1], Paediatric trainee; **Vivek Kalra[1],** Consultant Paediatrician.

Jenny Lind Children's Department, Norfolk and Norwich University Hospital, Norwich

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Introduction

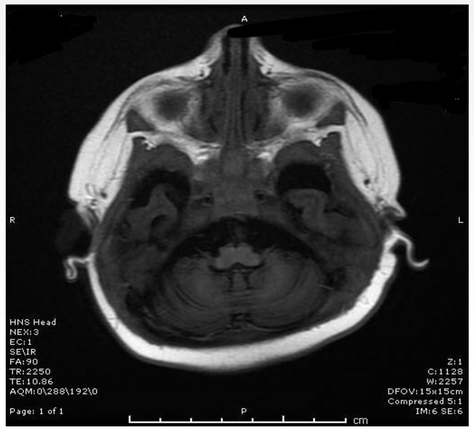
- Mutations in *SPTAN1* is a rare known cause for early onset epileptic encephalopathy.
- It presents with a distinct clinical syndrome. It is characterized by frequent severe refractory seizures and persistent abnormality of cortical function, which can be documented on electroencephalograms (EEGs). These features lead to impaired neurodevelopmental outcomes during neonatal or early infantile periods and beyond.1,2, 3.
- Additional early findings include; hypotonia, profound developmental delay with quadriplegia, acquired microcephaly, movement disorder such as opisthotonos posturing or dyskinetic movements and sever intellectual disability.
- MRI abnormalities include variable degree of atrophy affecting cortex, brainstem and/or cerebellum, hypomyelination, and thinning of the corpus callosum

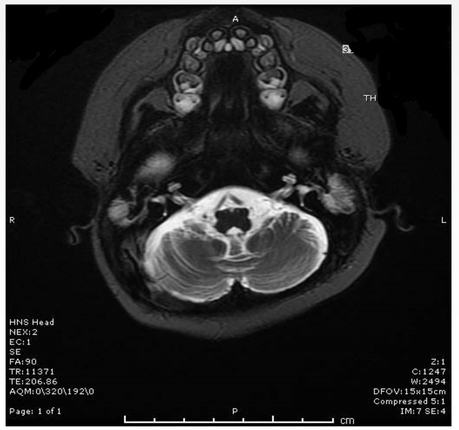
Objectives

To share a case study of a child diagnosed with SPTAN1 gene mutation and highlight peculiar features of this evolving distinct clinical syndrome.

Materials and Methods

- Our patient delivered at term in good condition following a normal pregnancy. She had poor tone, weak cry and choking episodes from early on.
- She presented at 3 months of age with infantile spasms and was treated with steroids and vigabatrin. After an initial response to treatment, she started having spasms refractory to treatment (Multiple AED's, ketogenic diet).
- **Examination** revealed marked hypotonia and Profound paucity of spontaneous movements.
- At 3 years, she showed significant global developmental delay, microcephaly, bilateral sensorineural deafness, nystagmus and showing signs consistent with evolving spastic quadriplegia with dystonic spasms.
- **Current medications**: Topiramate, Levetiracetam, Vigabatrin, Baclofen. Previously tried on - Clobazam, steroids, Ketogenic diet.
- An **EEG** revealed classical hypsarrhythmia with an earlier diagnosis of West syndrome. **MRI brain** showed cerebellar atrophy and subsequently she was found to have SPTAN1 gene mutation on WGS.
- TORCH screening, very long chain fatty acids, blood amino acids, ammonia, immunoglobulins, microarray, lactate, TFT, CK, U&Es, LFT, urine organic acids and acylcarnitine profile were done and came all normal.





MRI brain showing cerebellar atrophy

Conclusion

SPTAN1 encephalopathy has distinct clinical and neuroradiological phenotypes. Brainstem and cerebellar atrophy and cerebral hypomyelination, identified by MRI, are specific hallmarks of this disorder. Knowledge of clinical profiles and MRI features in *SPTAN1* encephalopathy will help clinicians to perform efficient genetic testing and to identify *SPTAN1* mutations

References

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