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CASE REPORT **OPEN ACCESS**

A 21-YEARS-OLD MAN WITH MESIAL TEMPORAL LOBE EPILEPSY AND DYSTONIA: A RARE CASE REPORT

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ABSTRACT

Background: Mesial temporal lobe epilepsy (MTLE) with dystonia is a rare case. Seizures and movement disorders have almost the same phenomenology, so it is often difficult to distinguish them. In this study, we report a unique case of MTLE and co-occurring dystonia. **Case:** A 21 years old male with complaints of seizures since 4 years ago. Seizures of one body jerking and drooling with a duration of less than 5 minutes. Prior to the seizure the patient was nauseous then vomited and followed by an empty mind, after the seizure the patient was confused. The patient also complained of unconscious movements in his right hand since 8 years ago. The movements disappeared when the patient slept. Physical examination revealed dystonic movement with a sensory trick on the right hand. Magnetic resonance imaging (MRI) of the brain with contrast showed bilateral hippocampal atrophy accompanied by left hippocampal sclerosis. Blood laboratory results, electroencephalography, and neurobehavior examination were within normal limits.. **Discussion:** MTLE can be caused by mutations in SCN1A, VPS13A, C90RF72, or TDP 43. Dystonia can be caused by mutations in SCN1A, TUBB4A, TOR1A, THAP1, or GNAL. SCN1A causes an increase in sodium influx, causing depolarization which causes clinical manifestations in the form of seizures and dystonia. For some disorders, although genetic causes have been identified, the molecular pathophysiology remains largely unknown, requiring further research. **Conclusion**: For some disorders, although genetic causes have been identified, the molecular pathophysiology remains largely unknown, requiring further research.

Keywords: dystonia; hippocampal sclerosis; mesial temporal lobe epilepsy; SCN1A

Introduction

Mesial temporal lobe epilepsy (MTLE) with dystonia is a rare case. Although they have similar clinical phenomenology and pathophysiology, they have different locations of lesions. The prevalence of MTLE with dystonia is very rare. The author has not found any previous case reports regarding MTLE with dystonia.

Movement disorders are a group of neurological conditions characterized by abnormal movements generally arising from altered function in the nuclei of the basal ganglia or their connections. Seizures are defined as the 'transient occurrence of signs and/or symptoms due to abnormal or excessive nerve activity in the brain, whereas epilepsy is a brain disease characterized by one or more seizures with a relatively high frequency of recurrence.[1]

The incidence of epilepsy in the world is 50.4 per 100,000 per year.[2] Mesial temporal lobe epilepsy (MTLE) is the most common form of epilepsy in adults.[2,3] Clinical manifestations of MTLE are 1) focal seizures, rarely developing into clinical tonic seizures; 2) the forms of aura that often occur are feelings of nausea, epigastric aura, psychic aura, for example

dejavu, and dreamlike states, followed by daze and automatism. Sometimes accompanied by autonomic symptoms such as vomiting. The awakening lasts 30 seconds to 2 minutes. The postictal state is characterized by disorientation in time and place, language disturbances when the dominant lobe is affected, coughing, and wiping the nose with the hand ipsilateral to the lesion. Seizures occur mainly when the patient is awake. The most common cause of MTLE is hippocampal sclerosis.[2] Dystonia is a syndrome of movement diorder in the form of persistent, contorted and repeated muscle contractions, causing abnormal postures. Dystonia is a subcategory of hyperkinetic movement disorders. Its prevalence is less than 1/2000 of the general population. The most common forms of isolated dystonia in adults are focal, affecting the neck (cervical dystonia), eyes (blepharospasm), or task-related (eg, writer's cramp).[4-8] So authors presented this case due to scarcity of relevant reports on seizures with movement disorders.

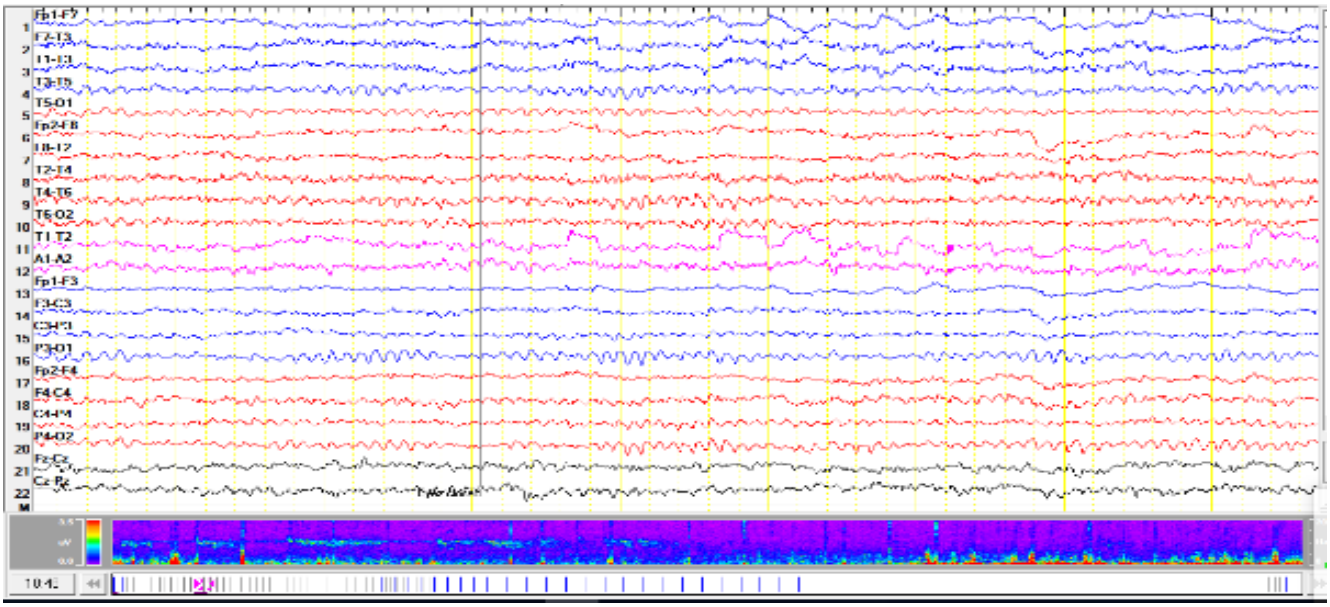
Case Report

A 21 years old man comes to the neurologist with complaints of seizures. Had the first seizure in April 2019. Before the seizure the patient felt nauseous then vomited and was followed by an empty mind. During the seizure the patient is unconscious, the whole body jerks, and salivation occurs, the duration of the seizure is less than 5 minutes. After the seizure the patient appears confused. The patient complained of the last seizure in January 2020. Since 2015, the patient has experienced involuntary movements in his right hand characterized by rough, irregular motions. These movements stiffen the patient's little finger upon onset and worsen with hand use, but improve when the hand is at rest or when supported by the left hand. No movements occur during sleep. Internal physical examination revealed no abnormalities. Neurologically, there were no deficits in cranial nerves, motor, sensory, coordination, or autonomic function. During extrapyramidal examination, dystonic movement with sensory trick was noted in the right hand.

Supporting examinations included blood tests, EEG (electroencephalogram), contrast-enhanced MRI (Magnetic Resonance Imaging) of the head, and neurobehavioral assessment. Kidney function, blood sugar, electrolytes, and thyroid function (Table 1), as well as EEG findings (Figure 1) and neurobehavioral assessment results (Table 2), were normal.

Table 1. Blood Laboratory Examination Results

Parameter	Results	Unit	Reference
Urea	20	Mg/dL	10-45
Creatinine	0.95	Mg/dL	0.50-1.10
Random blood sugar	128	Mg/dL	60-140
Sodium	145	mmol/L	135-145
Potassium	4.19	mmol/dL	3.50-5.50
Chloride	103.74	mmol/L	96-106
Ionic Calsium	1.25	mmol/L	1.10-1.35
TSH	3.420	uIU/mL	0.510-4.300
FT4	12.21	mmol/L	10.6-19.40



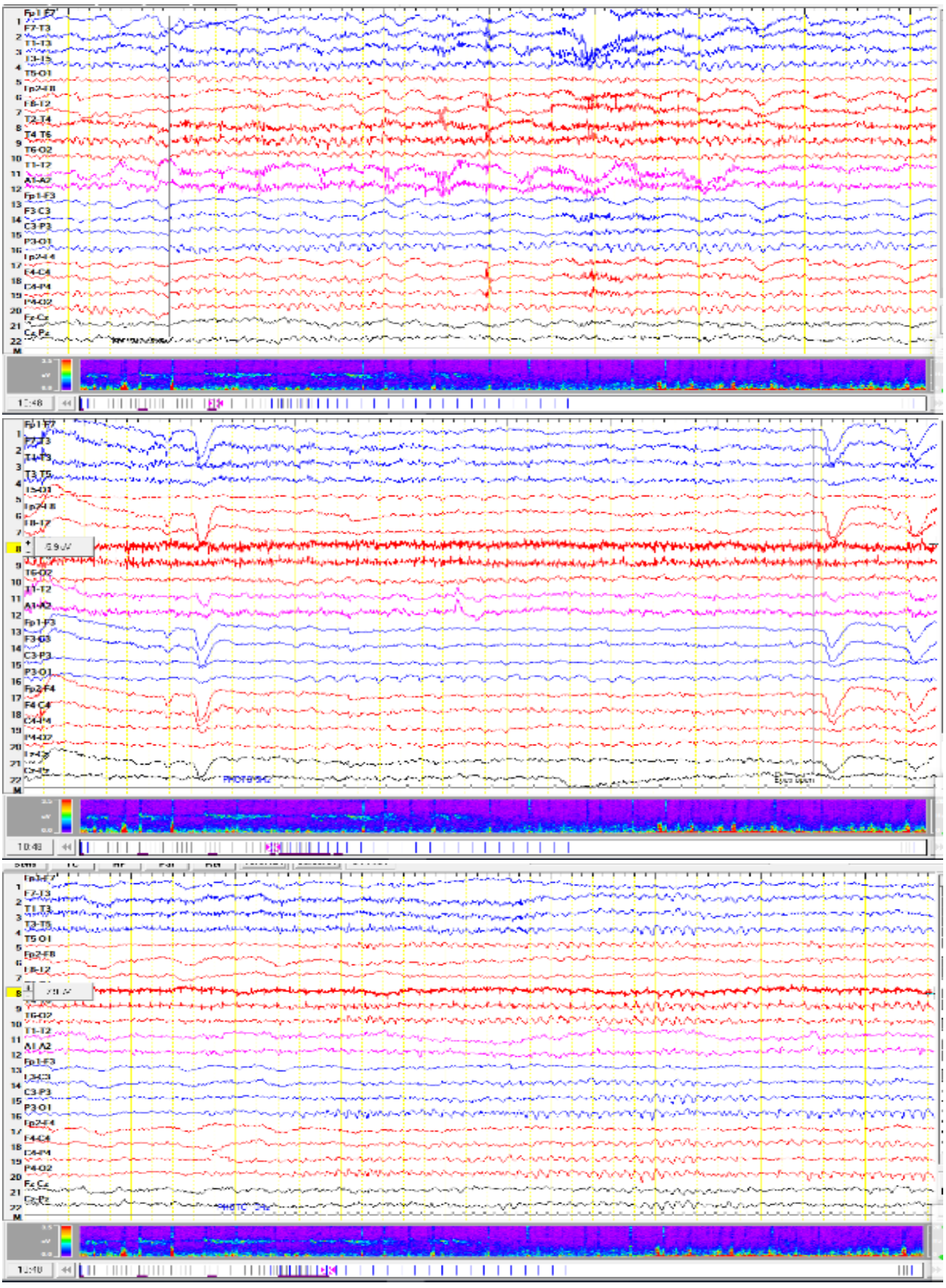
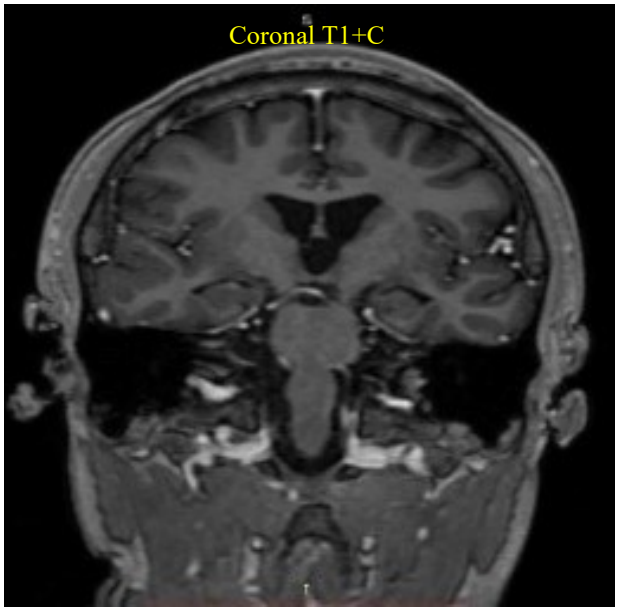
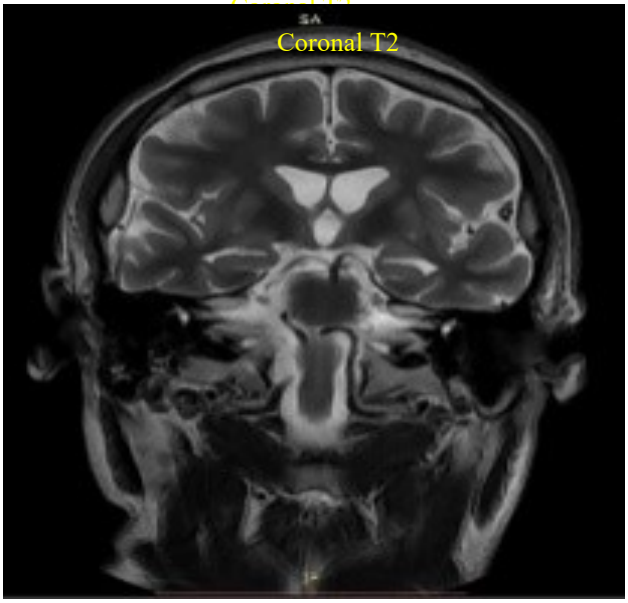


Figure 1. EEG results





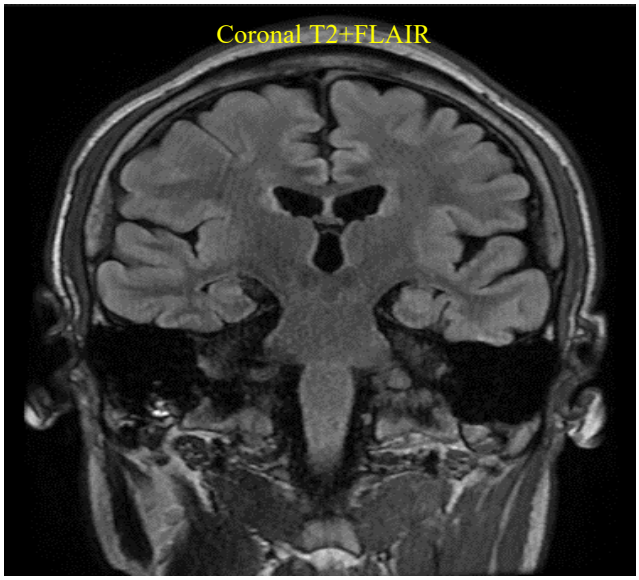


Figure 2. Head MRI Results with Contrast

However, upon conducting contrast-enhanced MRI of the head, significant findings revealed bilateral hippocampal atrophy characterized by notable reduction in volume and density, accompanied by mesial temporal sclerosis indicating a hardening of tissue in the mesial temporal region. Additionally, there was observed bilateral temporal lobe atrophy, characterized by significant shrinkage and loss of tissue integrity in these regions. These structural changes were quantified with an MTA score of II (Figure 2).

Table 2. Blood Laboratory Examination Results

No	Test	Score	Reference
1	Global Cognition Function		
	1.1 MoCA-Ina	28	≥ 26.7
2	Attention		
	2.1 Forward Digit Span	5	5
3	Language		
	3.1 Boston Name Test	13	13.84±1.23
	3.2 Verbal Fluency Test	19	19.84±5.98
4	Memory		
	4.1 Word List Memory Tasks	16	19.84±5.98
	4.2 Word List Memory Recall	7	8.05±1.66
	4.3 Word List Memory Recognition	19	9.88±0.36
	4.4 Recall of Construction Praxis	4	5.95±3.41
5	Executive Functions		
	5.1 Trail Making Test B	115	90.64±43.55

Discussion

Temporal lobe epilepsy consists of mesial temporal lobe epilepsy and lateral temporal lobe epilepsy. MTLE has the clinical specificity of focal seizures and an aura of nausea, epigastric aura, psychic aura, followed by gag and automatism, which is similar to this patient.

Temporal lobe epilepsy includes a variety of disorders that have a general picture of seizures arising in the temporal lobe. Pathologies that may underlie include tumors, vascular malformations, cortical dysplasia, trauma, and hippocampal sclerosis.[9] According to the theory, MTLE is most often caused by hippocampal sclerosis. The etiology of hippocampal sclerosis is due to genetic mutations SCN1A, VPS13A, PC90RF72, or TDP 43.[1,10-14]

Seizures can have multifactorial mechanisms, and they often appear so diverse that one would suspect that there is no common connotation. However, it is

commonly believed that seizures arise when the homeostatic mechanisms are disrupted, causing an imbalance between excitation and inhibition. Normally, there are checkpoints that keep neurons from excessive action potential discharging, and also mechanisms that facilitate neuronal fring so that the nervous system can function normally. Homeostatic disruption of the checkpoints or promotion of the mechanisms that enhance excitation can lead to seizures. Voltage-gated sodium channels are of great signifcance to the initiation of action potentials in neurons and other excitable cells, and their dysfunction causes epilepsy. Voltage-gated sodium channels function by transiently increasing the membrane permeability to sodium ions during membrane depolarization.[15] So if there is an error in the genetic coding of the sodium channel on chromosome 2q24.3, including SCN1A, can cause excessive depolarization and allow seizures to occur.[16]

Dystonia is a hyperkinetic movement disorder caused by genetic disorders, both familial and sporadic. There are several genetic mutations that affect dystonia, including mutations in TUBB4A, TOR1A, THAP1, or GNAL. This mutation plays a role in increasing sodium influx and decreasing GABA (gamma aminobutyric acid).[17,18] Brüggemann (2021) reported that to some degree, patients with dystonia exhibit temporal abnormalities. This abnormality can also be found in patients with genetic disorders.[19]

More recently, choreoathetosis, ballismus, dystonia, orofacial dyskinesia, stereotypies of the hand, and even cases of familial migraine hemiplegia have been described in patients with SCN1A.[20] The pathomechanisms causing these motor manifestations could lie in the function of the Nav1.1 and Nav1.2 channels (Voltage-gated Na+-permeable) which are both expressed in the basal ganglia.[16]

The weakness of this case report is the limitations in carrying out investigations to determine genetic abnormalities in patients, so the authors hypothesized that the pathophysiology in this patient was due to genetic mutations which resulted in increased sodium influx and decreased GABA.

Conclusion

The weakness of this case report is the limitations encountered in conducting investigations to determine genetic abnormalities in the patient. The authors suggest that genetic mutations causing increased sodium influx and decreased GABA levels may underlie the patient's pathophysiology. This hypothesis underscores the importance of further genetic research to elucidate the underlying mechanisms this condition.

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