eSLEEP EUROPE
October 4-6, 2023 | Virtual Congress

Abnormal sleep behaviors in a patient with 16p11.2 duplication syndrome – a diagnostic challenge

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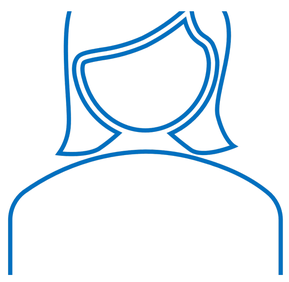
INTRODUCTION

- 16p11.2 duplication syndrome is a genetic condition associated with neuropsychiatric and neurodevelopment impairment.
- Although epileptic seizures are not uncommon in these patients, other non-epileptic events, such as sleep disorders, may occur. The heterogeneity in both abnormalities can contribute to challenging differential diagnoses.

CASE REPORT

IDENTIFICATION



Women, 20 years-old
16p11.2 duplication syndrome
Cognitive impairment
Frontal-central spike-wave during sleep in childhood
Medication: clobazam 10 mg; aripiprazol 15 mg

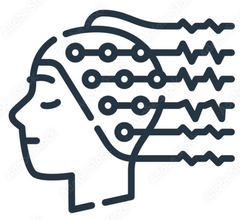
CLINICAL HISTORY

- Abnormal behaviors during sleep since childhood
- Frequency: weekly (1-2 per night), between 1-2 am
- Characterization: non stereotyped episodes with somniloquy/ screaming, or complex motor activity (raises trunk from the bed, claps, screams/talks), maximum 1min duration
- Absent dream or memory recall
- Absent snoring, morning headaches, nycturia, excessive daytime sleepiness (ESS=3)
- FLEP scale: -1

EXAMINATION

- BMI: 28.1 Kg/m2
- Examination: Mallampati: 2/4; auscultation: normal
- Neurological examination: puerile behavior, cognitive impairment, no focal deficits
- Brain MRI: normal

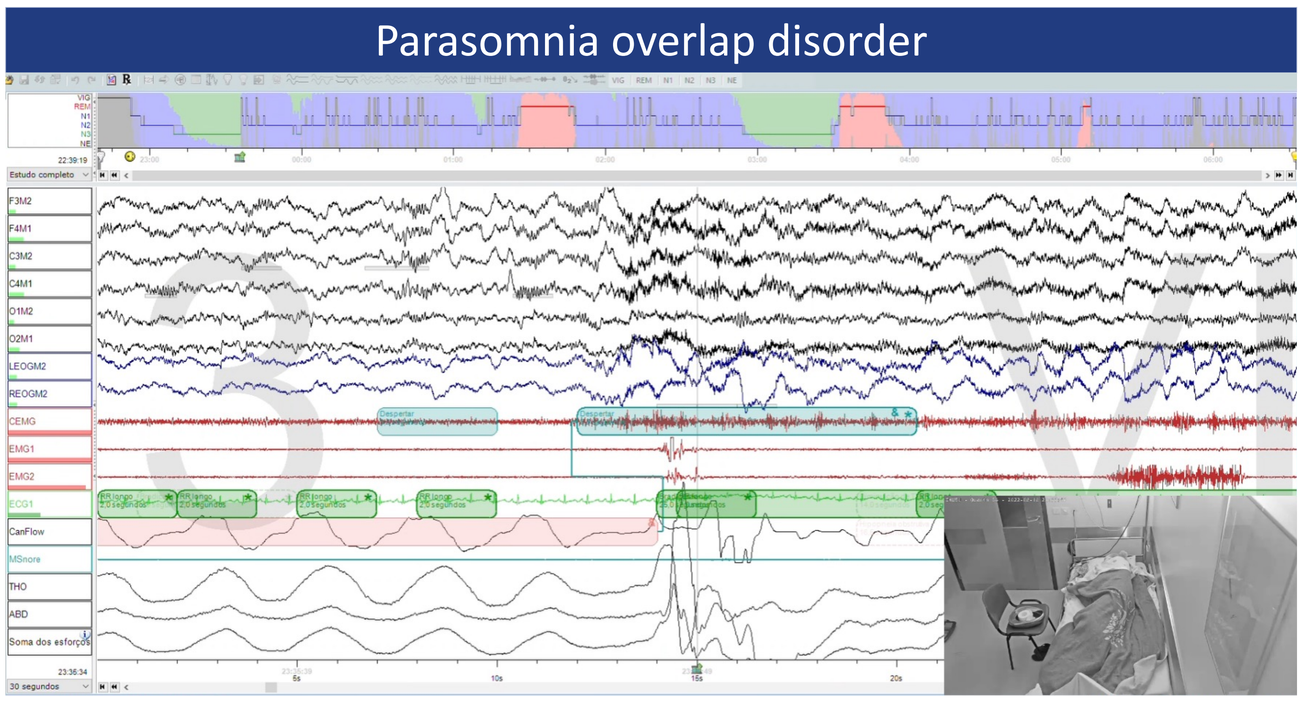
EEG



- 24-hour-VEEG: episodes of arousal from NREM (N2) and REM sleep, with non-stereotyped trunk or limb movements, and vocalizations; EEG with arousal, no epileptiform activity
- Interictal EEG: slow intermittent frontal-central activity, with no epileptiform activity

SLEEP STUDY

Parasomnia overlap disorder



Thirty-second PSG epoch with respiratory events (hypopneas) trigger arousal in N3 (figure and video) and REM (video), consistent with NREM and REM parasomnia; benzodiazepine suspension improved events

V-PSG:

- Moderate apnea/ hypopnea obstructive syndrome (AHI 15h/h) + sleep fragmentation

	Central Apnea	Obstructive Apnea	Mixed Apnea	Apnea (total)	Hypopnea	AHI
Number (n)	0	2	0	2	109	111
Index (n/h TST)	0.0	0.3	0.0	0.3	14.7	**15.0**

- Episodes of movements and vocalizations on N2 and N3 sleep, ***associated to respiratory events***
- 2 episodes on REM sleep, ***associated to respiratory events***

CONCLUSION

Parasomia overlap disorder can be potentiated by untreated OSA

Diagnosis in patients with genetic syndromes associated with epilepsy might be particularly challenging

Monitorings with VEEG and VPSG were important for excluding other causes, such as epilepsy and idiopathic parasomnias, whose treatment with benzodiazepines could worsen OSA.