ATR-X

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Background:

This study explores the prevalence, clinical characteristics, and treatment of epilepsy and sleep disorders in α thalassemia mental retardation (ATR-X) syndrome.

Design:

In this cross-sectional study, 37 participants with ATR-X syndrome aged 1.8 to 44 years were studied using a customized epilepsy questionnaire, review of electroencephalography (EEG) findings, the modified Sleep Questionnaire of Simonds and Parraga and 2-week sleep diary.

Results:

Eleven participants had a clinical diagnosis of generalized epilepsy (29.7%). Seizure types were generalized tonic-clonic seizures, absences, and myoclonia. Interictal EEG recordings in participants with GTCS showed no epileptic discharges in 78%. Similarly, EEG recordings during myoclonia and absences often demonstrated no epileptic discharges. Sleep problems (difficulty falling or maintaining sleep, and early awakening) were reported in 70%. Participants with reported sleep problems went to bed earlier ($p = 0.027$) and had a lower sleep efficiency ($p < 0.01$) than participants without sleep problems, but as a group they both had a sufficient total sleep time (9 hours and 52 minutes vs. 10 hours and 55 minutes). Sixteen participants (43.2) used medication to improve sleep (predominantly melatonin $n = 10$), being effective in only two.

Conclusion:

One-third of participants with ATR-X syndrome had a clinical diagnosis of epilepsy, but the absence of EEG abnormalities in suspected epileptic seizures questions this diagnosis in these patients. EEG recording during seizure like symptoms is warranted before making an epilepsy diagnosis. Seventy percent experienced sleep problems, although total sleep time was normal in most participants. Long bedtimes might have a negative influence on sleep efficiency.