Introduction

Background

Rett syndrome (RTT) is a neurodevelopmental disorder primarily caused by mutations in the MECP2 gene.

There are multiple health issues reported in RTT, including autonomic dysregulation and abnormal sleep patterns.

Previous studies have inconsistent findings when characterizing sleep/wake patterns in RTT, and have used a variety of methods to characterize sleep.

Purpose

This study aimed to replicate and expand upon previous work by examining multiple indicators of sleep using actigraphy, an objective measurement used to determine sleep/wake times, in addition to parent report in a sample of girls with RTT.

Methods

Participants and Setting

Following informed consent, thirteen Caucasian females with clinically diagnosed RTT participated in this study (M age = 9 years, range 1-17). All data were collected in the participants’ homes.

Methods

Caregivers were mailed a package containing a Philips Respironics Actigraph 2 device, a sleep diary to record their child’s bed time, night and daytime sleep, and wake up times, and questionnaires about each participant’s overall health and behavior.

The Actiwatch was programmed to collect data in 30 second epochs for seven consecutive 24 hour periods. A daily questionnaire accompanied the actiwatch data to document overall mood, alertness, additional medications taken, pain, and seizure activity for each day.

Analysis

There was a 9.5% actigraphy data loss (5 nights) due to non-compliance. Useable nights of actigraphy data ranged from 4-7 (Table 1).

The following variables were derived from actigraphy data using Philips Actiware 6 software: TNS: amount of time spent asleep while in bed; WASO: time spent awake after initial sleep onset; Sleep Efficiency: percent of time spent asleep while in bed; Daytime Sleep: time spent asleep when parents reported a nap.

Means and standard deviations were calculated for each sleep variable. Spearman correlations were run for individual participant comparisons of TNS (Table 1). A two-way random intraclass correlation was performed to compare parent reported TNS to actigraphy measured TNS. There were 78 out of 91 useable nights (85.7%) for this comparison, due to non-compliance.

Results

The sample had 553.5 mins (SD = 34.1) of mean TNS, 84.9% (SD = 6.3) sleep efficiency, 84.6 mins (SD = 36.8) of WASO, and mean daytime sleep of 47.2 mins (SD = 33.7) per day. All participants napped at least once. No age-related changes were observed for any sleep characteristic (Figure 2).

Comparison to Norms

Eight out of 13 participants (62%) had mean TNS values that fell within the National Sleep Foundation’s guidelines for recommended nighttime sleep. Seven girls (54%) had a mean sleep efficiency below 85%, a previously used cut off for poor sleep quality 4.

Parent Report vs. Actigraphy

Parents reported a mean of 48.4 four minutes (SD = 108.56) of TNS than measured by actigraphy. Parent report and actigraphy measured TNS were correlated (r = 0.56, F = 2.27, p < .01, df = 77, Figure 3), although correlation values differed substantially across participants (Table 1).

Discussion

Sleep patterns in RTT were variable across participants. Many of the participants met sleep guidelines for TNS, while sleep efficiency was poor for a majority of the sample. This indicates that some aspects of sleep/wake patterns of girls with RTT may be similar to those of typically developing children, while others may be atypical.

Expected age-related changes in TNS and daytime sleep were absent in the current study, replicating previous results. In typically-developing populations, TNS tends to decrease across the lifespan. Similarly, napping is rare after age 6 for typically-developing children, 5 in contrast with the current results. That said, the lack of an age-related change in sleep efficiency in this sample aligns with typically-developing patterns.

Although correlated, parents tended to over report TNS. This indicates that methodology of measuring sleep characteristics may influence results obtained, possibly explaining the inconsistency in the current literature of sleep in RTT. The variability in correlations across participants suggests that some parents may be more accurate reporters of their children’s sleep characteristics than others.

Limitations include a small sample size and a lack of a comparison sample. Future work will add more participants with RTT, as well as an age-matched, typically-developing control sample.

References