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SLEEP MEDICINE PEARLS

A Patient With Suspicious Oxygen Desaturations at Sleep Onset

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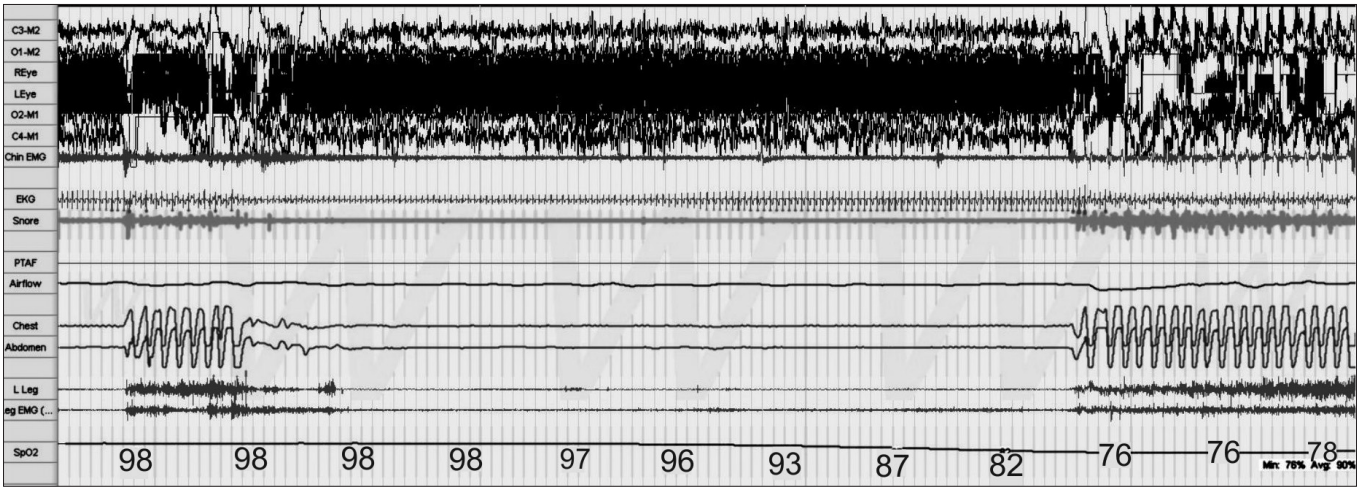
Your sleep tech asks you to check on the veracity of oxygen desaturations noted prior to sleep onset in a child who presented for a sleep study to evaluate the family's reports of apneic events. The patient is a 2-year-old female who was referred by a pediatric neurologist for further evaluation of the family's reports of apnea noted prior to sleep onset. The child was developing normally until approximately 6 months prior when she began to have regression of language and developed unusual hand movements. Physical examination was most remarkable for mild decrease in head circumference with child noted to be exhibiting wringing motions with her hands. You review the tracing shown in **Figure 1**. The event shown in **Figure 1** was noted during wakefulness without similar episodes noted during sleep. Polysomnography showed normal sleep architecture with an apnea-hypopnea index of 0.9 events/h.

QUESTION: What is the event depicted in the tracing and what is your diagnosis?

Figure 1—A 120-second tracing from a diagnostic polysomnography showing an event lasting 72 seconds with oxygen desaturation to 76% prior to sleep onset.



ANSWER: This tracing shows a prolonged central apnea prior to sleep onset, causing severe oxygen desaturation. This patient has Rett syndrome.

DISCUSSION

As pictured in **Figure 1**, this patient demonstrated a prolonged central apnea prior to sleep onset resulting in severe oxygen desaturation. This child has Rett syndrome which occurs in 1 in 8,500 live female births and is most often due to a mutation in the *MECP2* gene.[1] Most males with this mutation do not survive long after birth. This gene produces a protein that is important for normal brain function and development of normal brain synapses.[1] In most affected females, there is no family history of Rett syndrome with the mutation occurring de novo. There are a small number of familial cases reported with an X-linked dominant inheritance pattern. Children with Rett syndrome demonstrate normal development initially, and after age 12–18 months[2] they will then show lack of developmental progress with subsequent rapid loss of language and motor skills with loss of purposeful hand movements.[2] Many will have hand wringing or clapping with other signs/symptoms including seizures, hyperpnea, apnea, microcephaly, and tremor.

In children with Rett syndrome, breathing problems are often noted during wakefulness with periods of hyperpnea and subsequent central apneas. Marcus et al. examined polysomnography characteristics in 30 females with Rett syndrome and found a characteristic pattern of hyperventilation with subsequent central apnea and desaturation during wakefulness *not* noted during sleep.[3] No differences compared with a control group were noted in CO_2 values, number of obstructive events and number of central events with desaturation. Although arterial oxygen saturation during rapid eye movement sleep was slightly lower in patients with Rett syndrome (nadir 94% ± 2% versus 96% ± 2%) it remained within the normal range.[3] The authors suggested that this abnormal pattern was related to some form of cortical dysfunction.[3] Other sleep-related symptoms include laughing, bruxism, screaming, seizures and daytime napping.[4] Treatment is largely supportive, including physical and occupational therapy as well as treatment for seizures if needed.

SLEEP MEDICINE PEARLS

1. Children with Rett syndrome have a characteristic breathing pattern during wakefulness with hyperpnea followed by central apnea and desaturations. These episodes are not typically noted during sleep.
2. Rett syndrome should be considered in girls who present with developmental regression and hyperpnea/apneas noted during wakefulness. Male infants with the *MECP2* mutation often do not survive infancy.
3. Other sleep-related issues noted in Rett syndrome include daytime napping, nocturnal seizures, nocturnal screaming, laughing and bruxism.

CITATION

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