

# RETT SYNDROME AND SLEEP

2015

## FREQUENTLY ASKED QUESTIONS

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A good night's sleep promotes learning, improved mood, general good health, and a better quality of life for both your child and the whole family. This article written for [Rettsyndrome.org](http://Rettsyndrome.org) by Dr Daniel Glaze, Medical Director of The Blue Bird Circle Rett Center at Texas Children's Hospital and Sleep Disorder expert, reviews the benefits of sleep; the risks involved with disrupted sleep; the types of sleep problems experienced frequently in Rett syndrome; and strategies for getting back on track to good sleep habits. Your Frequently Asked Questions are answered!

### 1. WHY SLEEP?

In addition to diet and exercise, sleep is the third "pillar" contributing to a healthy life for children and adolescents and promoting the well being of the family.



### 2. HOW COMMON ARE SLEEP PROBLEMS IN CHILDREN AND ADOLESCENTS?

20-25% of typically developing children experience

some type of sleep disturbance.

It has been estimated that between 40% and 80% of children with neurodevelopmental disorders have sleep problems.

### 3. ARE CHILDREN/ADOLESCENTS WITH RETT SYNDROME (RTT) AT A HIGHER RISK FOR SLEEP PROBLEMS?

A large Australian study using the RTT database and questionnaires found sleep problems in over 80% of those with RTT. Nighttime laughing improved while daytime sleepiness worsened with age. Sleep problems were more severe with large deletions, and more common with p.R294X or p.R306C mutations. Similarly, fewer sleep problems were observed with p.R270X, p.R255X, and p.T158M mutations.

The Baylor College of Medicine RDCRN site has enrolled 284 individuals with RTT in the Rett syndrome Natural History Study. As part of their evaluation, questions concerning sleep problems were answered by their parents or care takers. Of the total group 30% were reported to have problems with initiation or maintenance of sleep, and difficulty arousing in the morning. Of the total group, 29.6% had problems going to sleep; 42.6% experienced frequent awakenings during the night, and 18.3% had difficulty arousing in the morning. There appears to be a significant age difference in the frequency of these problems. The youngest (< age 5 years; 35.9%) and the oldest (> age 20 years; 52.9%) RTT individuals were more likely to have problems with sleep initiation. Problems maintaining sleep were common in all age groups, while problems with morning arousal occur more frequently in the older patients (>Age 20; 35.3%). These findings emphasize that sleep problems occur frequently in RTT.

During the national meeting of the American Academy of Sleep Medicine we reported the findings characterizing sleep problems in children with rare genetic disorders. This study was performed in

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collaboration with the RDCRN and included children with Angelman syndrome, Rett syndrome and Prader-Willi syndrome, and the typically developing siblings of the children with these disorders. The findings of the study included the following:

- Sleep Problems occur frequently in Rett syndrome (84.3%), Angelman syndrome (91.4%) and Prader-Willi syndrome (64.6%)
- Sleep problems were common in the sibling control group (60.4%). This includes sleep problems in general as well as Sleep Disordered Breathing and Daytime Sleepiness.
- For the Rett participants clinical severity was related to daytime sleepiness but not to overall sleep problems or sleep disordered breathing. Mutational status showed no significant association to these problems.

The findings emphasize the importance of screening children with Rett syndrome and other neurodevelopmental disorders for sleep problems. The findings raise the possibility that the siblings of children with neurodevelopmental disorders in the household may be at an increased risk for sleep problems, though other factors including environmental and parenting skills and behaviors may also contribute.

### 4. ARE SLEEP PROBLEMS A COMMON PARENT COMPLAINT?

Sleep problems in children are the fifth leading concern after illness, feeding, behavior, physical abnormalities, and before language and motor development, toileting, and teething.

However, parents may not ask their physicians about sleep problems because they feel it is “normal” for children with Rett syndrome. Their physicians may not take the time to inquire about sleep problems. These beliefs and practices should change.

### 5. DO SLEEP PROBLEMS PERSIST?

If not treated sleep problems may become chronic. Several factors contribute to this and include neurodevelopmental delay; chronic illness including gastrointestinal problems; maternal depression; and, family stress. Some or all of these may play a role in the severity and persistence of sleep problems in Rett syndrome.

### 6. WHY IS SLEEP NECESSARY FOR CHILDREN'S OPTIMAL FUNCTION?

We don't actually know why we sleep. Popular theories suggest that sleep promotes restoration of brain cells and circuits; it may play a role in learning.

We do know the consequence of poor sleep and inadequate sleep. In general this leads to problems with mood like irritability; cognitive dysfunction including inattention and learning problems; behavior problems like impulsivity and over activity.

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### 7. WHAT IS THE IMPACT OF POOR SLEEP?

Sleep problems promote:

- Poor Mood regulation and Impulse control
- Poor Cognitive function
- Accidental injuries
- Compromises Cardiovascular (like high blood pressure), immune (resistance to infection) and endocrine (hormonal) function

### 8. DO SLEEP PROBLEMS HAVE AN IMPACT ON THE FAMILY?

In families with a child with sleep problems there is increased family stress and marital discord; poorer family sleep, daytime fatigue and job loss; and, decreased effective parenting, and potential for child physical abuse.

### 9. ARE PEDIATRIC SLEEP PROBLEMS TREATABLE?

Sleep problems are treatable. Treatment begins with identifying that children are experiencing sleep problems. However, of 600 community-based physicians: 20% did not screen for sleep problems; 25% did not screen for snoring; and, less than 40% questioned adolescents about sleep habits. A validated questionnaire developed for primary physicians is the BEARS. It consists of five developmentally appropriate screening questions concerning the following:

- B = Bedtime problems
- E = Excessive daytime sleepiness
- A = Awakenings during the night
- R = Regularity and duration of sleep
- S = Snoring

This questionnaire is a quick and efficient way to identify sleep problems in children. It yields more information than simply asking “does your child have problems sleeping”.

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### 10. HOW DO YOU EVALUATE SLEEP PROBLEMS IN CHILDREN WITH RETT SYNDROME?

Determine Parent Attitudes:

- How do the parents respond to the sleep problem?
- What is their goal/what works for them?
- How are the parents coping with their child's sleep problems?
- What is the impact of the child's sleep problems on family sleep and daytime activities?

Evaluate for medical problems that may contribute to sleep problems:

- Are there Gastrointestinal problems including gastroesophageal acid reflux (GERD)?
- Does she have Scoliosis which could contribute to breathing problems during sleep?
- Does your daughter have seizures, especially those occurring at night?

- Is your daughter taking medications—including those given for anxiety and behavior management, seizure medications, and over-the-counter sleep aids—which may have an impact on sleep?

Evaluate for sleep problems:

This begins with a sleep history to characterize difficulties initiating and maintaining sleep at night, unusual night spells and movements, and problems with breathing during sleep.

Your physician may ask you to complete a two week sleep diary for your daughter. You will be asked to record bed time, out of bed times, arousals, nocturnal events, sleep environment, bedtime routine, etc. This often reveals the sleep pattern including night time wakings and regularity of bedtimes, wake times, and naps.

The sleep diary below demonstrates that this child has a consistent bedtime, but does not go to sleep until 1-2 hours later. These findings suggest a delayed sleep phase.

**INFANT and CHILD SLEEP DIARY**

**TWO WEEK SLEEP DIARY FOR (Name) PAT**

1. Answer the questions in the shaded areas.
2. Draw a line through the times your child was asleep (include naps). Each box represents one hour.
3. Put down arrow (↓) at the times your child went to bed and up arrow (↑) at times your child got out of bed.

Date	Day	8:00 am	9:00 am	10:00 am	11:00 am	12:00 pm	1:00 pm	2:00 pm	3:00 pm	4:00 pm	5:00 pm	6:00 pm	7:00 pm	8:00 pm	9:00 pm	10:00 pm	11:00 pm	MIDNIGHT	Rating Scale:											
																			Rate your child's quality of sleep	Rate your child's level of quality of alertness	Rate your child's mood on awakening									
M	Day 1	X	X	X																↓	X	X	X	X	X	X	↑	2	1	1
T	Day 2		X	X	X															↓	X	X	X	X	X	X	↑	2	1	1
W	Day 3		X	X	X		X													↓	X	X	X	X	X	X	↑	2	1	1
Th	Day 4			X	X				X											↓	X	X	X	X	X	X	↑	2	1	1
F	Day 5		X		X			X												↓	X	X	X	X	X	X	↓	2	1	1
S	Day 6	X	X	X	X	X	X													↓	X	X	X	X	X	X	↓	3	2	2
S	Day 7	X	X		X		X	X												↓	X	X	X	X	X	X	↓	3	2	1
M	Day 8		↑		X		X			X										↓	X	X	X	X	X	X	↑	2	1	1
T	Day 9		X		X															↓	X	X	X	X	X	X	↓	2	1	1
W	Day 10		X	↑		X														↓	X	X	X	X	X	X	↓	2	1	1
Th	Day 11		↑		X		X													↓	X	X	X	X	X	X	↑	2	1	1
F	Day 12			X	X	X															↓	X	X	X	X	X	↓	3	1	1
S	Day 13	X	X	X	X	X	X														↓	X	X	X	X	X	↓	3	2	2
S	Day 14	X	X	↑	X	X		X												↓	X	X	X	X	X	↑	2	1	1	

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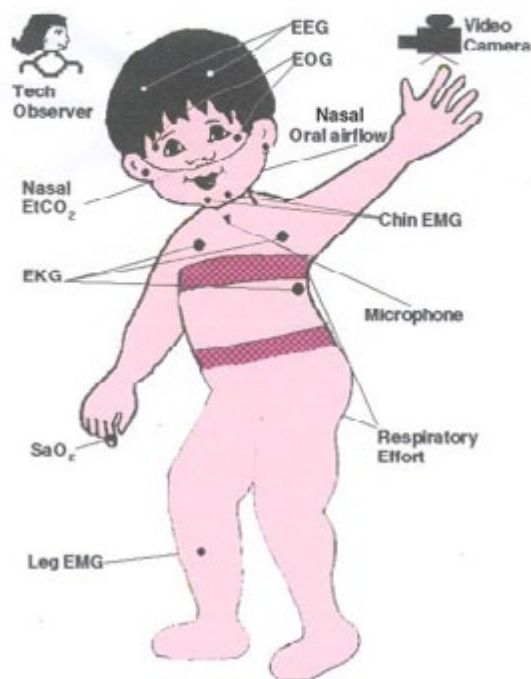
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For selected children an overnight sleep study may be performed: The sleep study includes the continuous recording of brain activity (EEG), airflow, breathing effort, oxygen and carbon dioxide levels, heart rate, and extremity movements. The sleep study should be done in a pediatric sleep laboratory, by a technologist (attended) and pediatric sleep physician, and include video taping. The study should run the whole night—the usual sleep period of the child. A parent stays with the child. This study is “non-invasive” and not painful, though placement of the sensors may require some restraint of the child.

In summary, a sleep study may be indicated if obstructive sleep apnea (OSA) is suspected; if the child is sleepy during the day in order to identify causes of nighttime sleep; to characterize nighttime behavioral episodes which may represent seizures or sleep phenomena such as night terrors or sleep related movements.

### INDICATIONS FOR PSG IN CHILDREN

- Suspected OSA
- Titrate CPAP
- Day Time Sleepiness
- Suspected Seizures/Injuries Nocturnal Spells
- Suspected PLMS



### 11. WHAT ARE THE MOST TYPICAL SLEEP PROBLEMS EXPERIENCED BY CHILDREN WITH RETT SYNDROME?

- Insomnia: Difficulty initiating and maintaining sleep; early morning awakenings
- Wake/sleep schedule problems: Delayed sleep onset; early morning awakenings; irregular schedules
- Sleep disordered breathing: Episodes of pauses in breathing including central sleep apnea (no effort and no airflow) and obstructive sleep apnea (effort to breathe but no airflow). These episodes occur during sleep and are different from the periods of breath-holding and hyperventilation that occur during wakefulness in Rett syndrome.
- Epilepsy: Some individuals with Rett syndrome experienced seizures only at night or more frequently at night in association with sleep.
- Parasomnias: These are unusual behaviors happening in relationship to sleep and may include so-called night terrors and episodes of laughing.

### 12. HOW DO YOU MANAGE SLEEP PROBLEMS IN CHILDREN WITH RETT SYNDROME?

#### Part 1

- Treat Medical Problems
- Manage Daytime Behavioral Problems
- Review and Modify Medications as Needed
- Provide Information and Establish Reasonable Goals for parents

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### Part 2

- Treat Other Sleep Problems (OSA)
- Sleep Hygiene
- Behavioral Approaches
- Medications

### 13. WHAT BEHAVIORAL TREATMENTS ARE USED TO TREAT SLEEP PROBLEMS?

- Extinction (“letting the child cry it out”); gradual extinction with progressively longer periods of non-response/ not reinforcing the behavior; extinction with the parent present (“the super-nanny method”. Gradual distancing yourself from the child)
- Scheduled Awakenings to anticipate the time of the undesired behavior
- Parent Education concerning sleep and appropriated expectations
- Relaxation: Massage Therapy

### 14. WHICH MEDICATIONS ARE USED TO TREAT SLEEP PROBLEMS IN CHILDREN?

- No Approved Medications for Sleep Problems in children
- Few or No Studies of Kinetics; Safety; Efficacy of Drugs
- However, Medications Frequently Used in children for sleep problems

Medications used to treat sleep problems frequently experienced by children

- Alpha Agonists: Clonidine (0.025-0.3 mg)
- Hypnotics: Zolpidem (5-10 mg; CR 6.25-11.5 mg)
- Antidepressant: Trazadone (25-50 mg)
- Melatonin (wide dose range; typically 3-5 mg; long acting CR 3 mg)

### 15. ARE SLEEP PROBLEMS PREVENTABLE?

Yes, by providing parents with information.

Sleep requirements reported for typically developing children:

- 0-2 months: 16-20 h; no night/day pattern
- 2-12 months: 9-12 h; 4-1 naps (2-5h)
- 1-3 yr: 12-13 h per 24 h; 2-1 naps

- 3-5 yr: 11-12 h per 24 hr; 1-0 naps
- 6-12 yr: 10-11 h; 0 naps
- 12-18 yr: 9-9.5 hr; 0 naps

Night wakings, when to be concerned:

- Frequency: > 3-4
- Duration: >20 minutes
- Require parent intervention
- Associated with Snoring; Movements; Injurious or Unusual (Stereotypic) Behaviors
- Daytime Sleepiness

What parents can do to promote good sleep:

#### SLEEP HYGIENE

- Age/Developmental Appropriate sleep schedule
- Nightly routine 20-30 Minutes Prior to Bedtime
- Put to Bed Awake but Drowsy
- Maintain regular Bedtimes and Nap Times
- Bed only for sleep; Bedroom should be Dark (night light may be used);
- Cool (<75 F); Safe; Electronic Media Free
- Pre-Bedtime Meal: Light Snack
- No Caffeine/Alcohol (many medications including OTC types contain alcohol)
- Routine Exercise: 3-4 Hours Prior to Bedtime
- Light: Bright light in AM Promotes Earlier Onset Sleep; PM Bright Light Delays Onset Sleep.

### FINAL THOUGHTS

Good sleep and adequate sleep promote good health and optimal functioning.

Every child, and their parents and family, deserves a good night's sleep.

Sleep problems occur commonly in individuals with Rett syndrome. Every visit to your child's primary physician should include an evaluation of sleep—if they don't ask; you should tell.

Sleep problems can be diagnosed and treated.

Sleep problems may be preventable or at least improved—be proactive.