**Difficult to Diagnose Epilepsy Cases**

*Children and Adolescents*

Lunluya Thampratankul, MD  
Division of Neurology, Department of Pediatrics  
Ramathibodi Hospital, Mahidol University  
Annual Meeting Epilepsy Society Thailand 2015

---

**Difficult to diagnose epilepsy cases**

- Epilepsy may be misdiagnosed in around 25% of cases.
- Conditions such as syncope, paroxysmal disorders or conversion disorder may be misdiagnosed as epilepsy.
- Alternatively, the symptoms of epileptic seizures may be misdiagnosed as resulting from psychiatric or associated disorders.

Smith D, Defalla BA, Chadwick DW. 1999.

---

**Difficult to diagnose epilepsy cases**

- The misdiagnosis of epilepsy may lead to human costs such as distress to patients and carers, unnecessary lifestyle changes, social stigma, social and financial deprivation as well as economic costs.

- The annual cost of epilepsy misdiagnosis in England is around £189 million.

All-Party Parliamentary Group on Epilepsy. The human and economic cost of epilepsy of epilepsy in England: wasted money, wasted lives. 2007

---

**Difficult to diagnose epilepsy cases**

People with intellectual disabilities are at additional risk of misdiagnosis:

- Stereotypical behaviors
- Drug induced involuntary movement
- Communication difficulties
- Dependence on the observation of carers
- Difficulty gaining an EEG
Cases:

• An 8 year-old boy with nocturnal spells and abnormal EEG

• A 17 year-old male with congenital heart disease, epilepsy and new onset of nocturnal spells

• A 2-year-old boy with tongue biting during sleep

An 8 year-old boy

Chief complaint: nocturnal spells for one year.

Present illness:
12 months PTA: While being hospitalized for high grade fever, he has developed confusional episode during sleep which occurred every night.
• 30-40 minutes after falling asleep, he would get up, scream inconstantly, try to climb down from his bed and occasionally escape from his bedroom.

Present illness (cont.)

• These episodes last anywhere from 5 – 30 minutes. They occur every night, about 1-2 times each night and may be worsened after tiredness (up to 3 times).

• He could not recall any episode and there is no difficulty in waking him up in the morning.

• Additionally, he snores every night and is very restless in his sleep since he was 3-year-old.

Present illness (cont.)

• The mother brought him to a hospital where further investigations were performed.

  ❖ EEG: frequent spikes over the right centro-parietal regions
  ❖ MRI brain: normal

• Levetiracetam and subsequently diazepam were given, but neither improved his symptoms.

Sleep history

• Sleep time: 8 PM
  • Wake up time: 6-7 AM
  • Denies daytime nap or daytime somnolence
  • (+) nocturnal enuresis 2 time/week
  • (+) snoring, restless sleep
  • Bedroom: sharing with parents and one brother
Past history
- Allergic rhinitis
- Otherwise unremarkable

Family history
- (+) sleep walking in mother, disappeared after age of 10 years
- (-) seizure or other neurological condition

Physical Examination
- Alert, well cooperative
- BT 37 °C, BP 95/55 mmHg, HR 80/min, RR 20/min
- BW 26 Kg (P75), Height 125 cm (P50)
- HEENT: swollen and pale inferior nasal turbinate, tonsils 3+ both sides, Mallampati class III
- Heart, lungs, abdomen: normal
- No focal neurological deficit

Problem List
- Epileptic or non-epileptic event?
  - Nocturnal spells
  - Abnormal EEG (Right centro-parietal spikes)
- R/O obstructive sleep apnea

Problem List
- Allergic rhinitis
- Snoring
- Enlarged tonsils
- Restless sleep
- R/O obstructive sleep apnea

Problem List
- Epileptic seizure or Parasomnia?
  - R/O obstructive sleep apnea

An 8-year-old boy with “Nocturnal Spells”
- Early phase of sleep (First 1/3 of the night)
- Non-stereotype
- Family History of parasomnia
  - Abnormal EEG

Provisional Diagnosis
- Non REM parasomnia with suspected OSA
Differential Dx:
- Nocturnal seizures
**Hypnogram:**
Events occurred in slow-wave sleep

**Polysomnography with Extended EEG**
- Sleep efficiency: 50.64%
- N1 5%, N2 52%, N3 38%, REM 5%
- Apnea-Hypopnea Index (AHI): 1.2/hr (REM index 8.1)
- Low sleep efficiency
- Decreased REM sleep
- Low AHI in total sleep, but high in REM.
- This study may underestimate severity of OSA due to inadequate REM sleep.
- EEG: frequent independent spikes over left and right centro-parietal regions
- Abnormal EEG, but not correlate with the events.

**Diagnosis**
- Low sleep efficiency (first night effect)
- Mild OSA
  - Allergic rhinitis, tonsillar hypertrophy
- Non-REM parasomnia

**Treatment**
- Reassure, discontinue AEDs
- Rx AR and mild OSA with intranasal steroid
- Rx parasomnia

**Management of Parasomnia**
- REASSURE
  - Sleep hygiene:
    - regular sleep-wake cycle
    - adequate amount of sleep
  - Manage triggering factors
    - Avoid all stimuli that may contribute to partial arousal and trigger an episode e.g. noise, light
    - Avoid extreme exercise, emotional or situational stress
    - Avoid antihistamine, alcohol, antidepressant, sedative
  - Environmental safety
    - Search for and treat, if present, other sleep disorder, such as OSA, PLMD, RLS
    - Scheduled awakening to eliminate sleep walking
    - Clonazepam for recurrent and problematic parasomnia

**Case Demonstration:**
- An 8 year-old boy with nocturnal spells and abnormal EEG
- A 17 year-old male with congenital heart disease, epilepsy and new onset of nocturnal spells
- A 2-year-old boy with tongue biting during sleep
**A 17-year-old male**

**Chief complaint:** Unusual behavior during sleep for 3 months

**Present illness:**
- Known case hypoplastic left heart with pulmonary atresia s/p Fontan operation
- **17 years PTA:** At age of one month, he was diagnosed with meningitis due to fever with seizure and was put on phenobarbital.

**Present illness (cont.)**

His seizures were very well controlled and the medication was discontinued at aged of 3 years.

- **10 years PTA** (after 6+ years of no medication): He had recurrent seizures, characterized by clonic movement of left side of face and left arm. Sodium valproate was given.
- **3 years PTA:** After 7 years of seizure free and normal EEG, VPA was slowly discontinued.

**Present illness (cont.)**

**6 months PTA:** He had loud snoring and occasional grasping without any witnessed apnea. His weight increased 14 kg in 2 years.

- **3 months PTA:** He has had nocturnal spells which were not similar to his previous seizures. “restlessness, body turning lasting 10-30 seconds” These occurred 1-3 times every night. After the episode, he might wake up or return back to his sleep without any recall.

**Sleep History**

- Sleep time: 8 PM
- Wake up time: 5.30-6 AM
- Denies daytime nap, daytime somnolence
- Denies morning headache
- Bedroom: sleep in the same room with parents

**Past History**

- Congenital heart disease s/p surgery at age of 2 and 7 mo, currently on warfarin and captopril
- Intellectual disability, IQ = 76
- Otherwise are unremarkable

**Family History**

- (-) epilepsy, parasomnia
- Unremarkable

**Physical Examination**

- Alert, well cooperative, follow simple command appropriately, obese
- BT 37 °C, BP 128/64 mmHg, HR 86/min, RR 20/min
- BW 66 Kg, Height 156 cm, BMI 27.1 kg/m²
- HEENT: Not pale, anicteric, short neck, acantosis nigricans, tonsils 1+ both sides, Mallampati class IV
- No focal neurological deficit except for intellectual disability
Problem List

Hypoplastic left heart syndrome with PA s/p surgery
Post-meningitis with recurrent epilepsy, off AED
Obesity and snoring R/O OSA
Nocturnal spells

Diagnosis of Nocturnal Spells

• Brief spells, stereotype
• Variable time across the night
• Snoring and obesity R/O OSA

Differential diagnosis
Secondary parasomnia
(confusional arousal from OSA)
Nocturnal seizure

Overnight Video-EEG/PSG

EEG:
• Interictal: few spikes over left an right frontal regions (Fp1, Fp2).
• Ictal: 3 habitual spells captured, consistent with frontal lobe seizure

PSG:
• Cannot perform respiratory monitor due to non-cooperation

Final Diagnosis:
Nocturnal frontal lobe epilepsy

Management
• Control seizure with topiramate
• Diet control and weight reduction

Parasomnias vs Epilepsy
**Comparison of Clinical and video-EEG/PSG of Parasomnia and Nocturnal Frontal Lobe Epilepsy**

<table>
<thead>
<tr>
<th>NREM parasomnia</th>
<th>NFLE</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age at onset</strong></td>
<td>Usually &lt; 10 yrs</td>
</tr>
<tr>
<td>(+) Family Hx</td>
<td>60-90%</td>
</tr>
<tr>
<td><strong>Attacks per night</strong></td>
<td>1 or 2</td>
</tr>
<tr>
<td><strong>Episode per month</strong></td>
<td>&lt; 1 - 4</td>
</tr>
<tr>
<td><strong>Clinical course</strong></td>
<td>Tends to disappear by adolescence</td>
</tr>
<tr>
<td><strong>Episode duration</strong></td>
<td>Seconds to 30 min</td>
</tr>
<tr>
<td><strong>Semiology of movement</strong></td>
<td>Variable complexity, not stereotyped</td>
</tr>
</tbody>
</table>

Derry CP, et al. Epilepsia 2006

---

**Case Demonstration:**

- An 8 year-old boy with nocturnal spells and abnormal EEG
- A 17 year-old male with congenital heart disease, epilepsy and new onset of nocturnal spells
- A 2 year-old boy with tongue biting during sleep

---

**A 2-year-old boy**

**Chief complaint:** Tongue biting during sleep for 6 mo

**Present illness:**

**6 months PTA:** The mother has noted that he bit his tongue during sleep both days and nights. It occurs every day, several times per night. Afterward he will occasionally wake up and complain of pain in his mouth. He has ulcers at tips and on both sides of his tongue.

---

**Present illness (cont.):**

- This mainly occurs first half of the night and occasionally later half.
- The mother denies any seizure or jerky movement.
- He was seen by a dentist and “close bite” was diagnosed. He was put on mouth guard at night but his symptoms did not get better.
- No other self-injurious behavior.

---

**Sleep history:**

- Sleep time: 8 PM
- Wake up time: 6-7 AM
- Daytime nap 2-3 hours/day
- Denies daytime somnolence, snoring
- (+) restless sleep, sleep talking
- Bedroom: sleep in the same room with parents
Past History

- Unremarkable
- Fullterm, no perinatal complication
- Normal development for age

Family History

- (-) seizure, parasomnia or other neurological condition

Physical Examination

- Alert, well cooperative
- BT 37 °C, BP 95/55 mmHg, HR 80/min, RR 20/min
- BW 11.6 Kg (P75), Height 86.3 cm (P50)
- HEENT: tonsils 2+ both sides, Mallampati class III, old ulcers at tip of tongue.
- Heart, lungs, abdomen: normal
- No focal neurological deficit

Differential Dx of Tongue Biting During Sleep

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Sleep stage</th>
<th>Symptoms</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>Nocturnal seizure</td>
<td>Any, N2*</td>
<td>Hypermotor, stereotype</td>
<td>Vaznikov P, 1997</td>
</tr>
<tr>
<td>Rhythmic movement disorder of sleep</td>
<td>Sleep onset</td>
<td>Gradually disappear when older, may have head baring, rocking</td>
<td>Vaznikov P, 1997</td>
</tr>
<tr>
<td>Sleep bruxism</td>
<td>N1, N2</td>
<td>Rhythmic involuntary contractions of masticatory muscles, teeth grinding</td>
<td>International classification of sleep disorder 2nd</td>
</tr>
</tbody>
</table>

Differential Dx of Tongue biting during sleep

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Sleep stage</th>
<th>Symptoms</th>
<th>Reference</th>
</tr>
</thead>
<tbody>
<tr>
<td>Geniospasm</td>
<td>Awake, sleep (N2)</td>
<td>Decreased in REM</td>
<td>Sporadic, AD, infantile onset, increased with stress</td>
</tr>
<tr>
<td>Psychogenic</td>
<td>Awake</td>
<td></td>
<td>Jarman PR, 1997</td>
</tr>
</tbody>
</table>

Take home message

- To evaluate nocturnal event, a careful history is needed.
  - Time of sleep-wake occurrence
  - Precipitating factors: emotions, sleep deprivation, tiredness
  - Duration and frequency of episodes
  - Other accompanying features: pallor, diaphoresis, papillary dilatation, posturing
  - Factors associated with cessation or following the event
  - Documentation of the episode with home video recording**
  - Video-EEG/PSG recording in selected cases

Thank you for your attention