Central sleep apnea: case studies.

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Congenital Central Hypoventilation Syndrome in Children

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**Table 1**
Differential diagnoses of Congenital Central Hypoventilation Syndrome

<table>
<thead>
<tr>
<th>Metabolic:</th>
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<tbody>
<tr>
<td>Mitochondrial defects e.g. Leigh's disease</td>
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<td>Pyruvate dehydrogenase deficiency</td>
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<td>Hypothyroidism</td>
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<th>Neurologic:</th>
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<tr>
<td>Structural central nervous system abnormalities e.g. Arnold Chiari malformation, Moebius syndrome</td>
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<td>Vascular injury e.g. CNS hemorrhage, infarct</td>
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<td>Trauma</td>
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<tr>
<td>Tumor</td>
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<th>Pulmonary:</th>
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<tr>
<td>Primary lung disease</td>
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<td>Respiratory muscle weakness e.g. diaphragm paralysis, congenital myopathy,</td>
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<th>Genetic:</th>
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<tr>
<td>Prader Willi Syndrome</td>
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<td>Familial dysautonomia</td>
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<th>Sedative drugs</th>
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Causes

+ 

- Obesity
- Obstructive sleep apnea
- Idiopathic central sleep apnea
Case 1

• Boy, 8 years old.
• Referred for snoring and witnessed apneas.
• Past medical history:
  - Adenotonsillectomy
  - Respiratory allergies – momethasone spray, ceterizine
  - Chronic headache – no causes identified at a local hospital including brain MRI

• Investigations?
Case 1

• Breathing pattern?
  - CAI 111; 35% of TST with periodic breathing
• Investigations?
• Treatment?
Case 1

- Cardiorespiratory status:
  - Normal chest X-ray
  - Normal spirometry and lung volumes
  - Normal cardiac ultrasound

- Negative toxicology screen

- Negative PHOX2B analysis

- Respiratory alkalosis on blood gas
Case 1

- Respiratory alkalosis on blood gas
- Normal liver and kidney function
- Normal thyroid panel
Case 1

- **Management?**

  - Acetazolamide

  - **Non-invasive ventilation:**
    - Settings?
    - Trilogy - S/T
    - IPAP 9 cm H2O – EPAP 5 cm H2O – back up rate 14 per minute

    - Normal oximetry recordings at home – 60% triggering of breaths by the ventilator
Case 1

- New evaluation of the original MRI: Arnold-Chiari type I

- Posterior fossa decompression
  - CAI 111; 35% of TST with periodic breathing
  - 3 months after surgery: CAI 10, mainly associated with arousal and mild desaturations > 90%
  - 9 months after surgery: CAI 1.6
Case 2

- 2,5 year old boy with Prader-Willi.
- Severly obese. Short stature.
- Mental retardation.
- Diagnostic polysomnography because of snoring:
  - Obstructive AHI 23
  - Mean SaO2 95.4 %
  - 4 episodes of periodic breathing with less severe desaturation.
  - Heavy snoring with flow limitation.
Case 2

• Conclusion: Severe OSA.
• ENT: Massive adenoids and tonsillar hypertrophy.

• Adenotonsillectomy:
  - Hypoxemia improved but did not normalize over the next few days.
Case 2

- Polysomnography after three months:
  - Obstructive AHI = 14, mainly during REM.
  - Mean SaO2 93%.
  - No snoring.
  - Chronic rhinitis.

- Conclusion: residual moderate-to-severe OSA after adenotonsillectomy.
  - CPAP/NIV titration.
Case 2

- Treatment for chronic rhinitis was started (antibiotics for 1 week, ceterizine and flixonase).
- CPAP:
  - No abnormalities during NREM.
  - Central and mixed events during REM with high CPAP pressures.
- Bilevel ventilation:
  - Started on IPAP 8, EPAP 4, back-up rate 12.
  - Better during REM sleep, not normal.
  - Patient did not tolerate higher pressures.
Case 2

- Patient was sent home on these low bilevel settings to get used to the machine, but did not tolerate sleeping with the mask.
- Chronic rhinitis was resolved.
- Control polysomnography showed the presence of central apneas and periodic breathing during REM with persistent oxygen desaturations. No obstructive events were noted.

- Management?
Case 2

- The patient was started on acetazolamide 250 mg.

- Control polysomnography after 4 days of treatment:
  - Better baseline saturation. Mean SaO2 increased to 95%.
  - Less central events.
  - Less severe oxygen desaturation during REM.
Case 2

- After growth hormone treatment:
  - Mixed sleep apnea with again mainly obstructive events with oAHI 43; <SaO2> 92%, SaO2nadir = 61% and mild hypercapnia (CO2 of 49 in the morning).

  - Management – intolerant for NIV?

  - Supplemental oxygen 0.3 – 0.5 lpm: oAHI 11; <SaO2> = 96.7%, SaO2nadir = 70% and mild hypercapnia (CO2 of 47 in the morning).
Case 3

- 9 year old boy with Pitt-Hopkins syndrome who developed serious attacks of hyperventilation followed by apnea and syncope since the age of 7 years. These episodes occurred on a daily basis.
- MRI CNS: signs of cerebral atrophy, a normal aspect of the brain stem and cerebellum and no evidence of a Chiari malformation.
- No epilepsy.
Case 3

• Management?
Case 3

- Started on acetazolamide 250 mg once daily.
- Immediate clinical improvement: long apneas and syncope were no longer observed.
- Blood gas: pH 7.35; pCO₂ 32.9; BE -6.5.
Case 3

- Acetazolamide was stopped after 5 months. Hyperventilation and longer apneas reappeared.

- Acetazolamide was started again. Intermittent hyperventilation and longer apneas.
Case 4

- Girl, 10 months old.
- Presents with ALTE.
- Snoring during colds.
- No other symptoms.

Past medical history:

- Term delivery
- Neonatal sepsis with E. coli
- Recurrent otitis
Case 4

- Polysomnography:
  - TIB 655 min.; TST 530 min.
  - Sleep latency 0 min. SOREM 6.1 min.
  - Normal EEG. Het eeg was normaal.
  - Stage 1 3%; stage 2 21%; stage 3 32%; REM 39%
  - oAHI 20.8. Total AHI 27.8. No snoring.
  - $\langle SaO2 \rangle$ 96.3%; SaO2nadir 78%.
  - Arousal index 21.
  - Breath holding spells when applying electrodes.

- Diagnosis and management?
Case 4

- ENT evaluation – upper and lower airway endoscopy
  - Mild hypertrophy of the adenoids but not obstructive
  - No candidate for ENT surgery

- Home cardiorespiratory monitor with frequent alarms

- Management?
Case 4

- **CPAP**
  - CPAP 4 cm H2O: immediate occurrence of central apneas and periodic breathing
  - oAHI 3; total AHI 12; 13% of TST with periodic breathing
  - Meanwhile, patient developed serious constipation

- **Management?**
Case 4

- All results came back normal

  - No Hirschsprung disease

  - Idiopathic central sleep apnea:

    - started on acetazolamide 6-8 mg/kg
    - No alarms on cardiorespiratory home monitoring
    - Normal oximetry
    - Normal polysomnography after 6 months (acetazolamide was stopped 1 month before PSG)