

Sleep and Neuromuscular Disease

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Case Presentation Part I

- GR is a 21-year old male with Becker muscular dystrophy who comes to your office complaining of progressively worsening shortness of breath at rest and with exertion for the past year. His muscle strength has been slowly weakening over the past year as well, but he continues to be ambulatory and not wheelchair bound.
- His mother accompanies him to the visit and reports that he has been more tired recently and gets winded easily with little exertion. They have checked his oxygen saturation at rest and it is usually > 95%.

Questions to Consider

- What are important components of a good pulmonary / sleep history for patients with neuromuscular disease?
- What measures of pulmonary function are important in this patient population?
- What questions can be used to better quantify the risk of worsening pulmonary function?

Case presentation

- Upon further questioning, GR reports that his baseline pulmonary function tests (PFTs) have declined to a vital capacity of ~40% of predicted. He uses volume recruitment and deep lung inflation using a self-inflating manual ventilation bag twice daily. He has not developed any respiratory infections, and his baseline peak cough flow is > 270L/min. He also reports that he wakes up frequently at night and does not feel well-rested in the mornings. He also is feeling more tired and lethargic during the daytime.

Case presentation

- His physical exam is remarkable for blood pressure of 125/75, oxygen saturation of 96% on room air, body mass index of 25kg/m², Mallampati score of 2, and neck size of 16.5 inches. His respiratory exam reveals decreased diaphragmatic excursion. His cardiac, and abdominal exams are normal. He walks upright with a normal gait. He has 4/5 strength in the bilateral lower extremities and 2+ pitting edema of the bilateral lower extremities.

Questions

- What are the important components of a good physical exam in patients with neuromuscular disease?
- What are symptoms that may point to declining pulmonary function?
- What is the differential for shortness of breath in this population?
- When and how often should you obtain PFTs in this population?

Part II: Diagnostic Testing

- You decide that GR is high risk for pulmonary complications from his neuromuscular disease, and obtain PFTs.
 - PFTs: FEV1 2.03L (88%), FVC 2.26L (69%), FEV1/FVC ratio 90, VC 2.13L (65%), TLC 3.88L (76%), MIP -50cmH₂O, MEP 75cmH₂O
 - 6MWT: Walked 450m with desaturation to 88% and increased heart rate to 130bpm
 - ABG: 7.40/56/98/34

Questions to Consider

- When are manual and mechanically assisted cough techniques recommended in this population?
- When should we consider nocturnal ventilatory assistance?
- What tests could be ordered to evaluate the patient's need for nocturnal ventilation?

Part III: Diagnostic Testing

- You are concerned regarding nocturnal hypoventilation and hypoxemia and request an in-lab attended polysomnogram. The sleep study report is as follows:
 - Sleep Architecture: Exam started at 2022 and ended at 0440. Sleep latency was 25mins, and REM latency was 90mins. Sleep efficiency was 85%. Patient had normal distribution of stage N1, N2, N3, REM. He had 236 arousals during the exam.
 - Respiration: AHI was 15/hr and there was evidence of hypoventilation with the average event lasting 33 seconds with a maximum duration of 62 seconds. Episodes of hypoventilation were worse during supine REM.
 - Oxyhemoglobin saturation: Mean oxyhemoglobin saturation was 96%. Oxyhemoglobin saturation was below 88% for 20 minutes. Nadir oxygen saturation during episodes of hypoventilation was 75% on room air.

Questions to ponder

- What tests are available to diagnose hypoventilation syndrome associated with neuromuscular disease?
- How should sleep disordered breathing be treated in this population?

Part IV: Treatment

- Based on the results of the polysomnogram, you make treatment recommendations.

Questions

- What treatment options are available for nocturnal hypoventilation and hypoxemia in neuromuscular patients?
- What precautions have to be taken with use of NIPPV in these patients?

Treatment

- After some discussion GR accepts your recommendations and opts to use AVAPS
- You arrange for an in-lab AVAPS titration to ensure that he is titrated to comfort and doesn't have a mask leak

Treatment

- GR returns 1 year later – he continues to use nocturnal AVAPS and feels like his sleep has improved. However, he complains that he thinks his disease has progressed and he is now getting breathless just with talking.
- On exam, his oxygen saturation at rest is 91%.

Questions

- When would you recommend daytime ventilation?
- What are the guidelines regarding tracheostomy placement in these patients?