LEARNING OBJECTIVES

➢ To consider neurologic etiologies when evaluating a patient with respiratory failure.
➢ To recognize a common presentation of an uncommon neuromuscular disease.
➢ To contemplate a unifying etiology when presented with multi-system disease.

CASE PRESENTATION

➢ A fifty-four-year old woman presents for perioperative evaluation prior to hysterectomy and oopherectomy for grade one endometrial cancer.

- Over the past five years, she has had generalized weakness with recurrent falls and now uses a wheelchair and walker.
- More recently, she has developed increasing somnolence, sleeping twenty hours daily.

➢ Past Medical History
  - Developmental delay with mild speech impediment
  - Adult-onset diabetes with retinopathy
  - Hypertension with associated cardiomyopathy
  - Bifascicular conduction block
  - Ischemic stroke vs subarachnoid hemorrhage with residual left-sided hemiparesis
  - Iron deficiency anemia
  - Bilateral cataracts

➢ Family History
  reveals members with developmental delay and premature death from cardiomyopathy.

➢ Significant Physical Exam Findings
  - Long face with relative microcephaly, dental overbite and crowding of teeth
  - Bilateral ptosis and diminished abduction/adduction of both eyes with inability to completely close either eye
  - 1/6 systolic ejection murmur at the right upper sternal border
  - Distal upper/lower extremity weakness with myotonic hand grip
  - Bilateral foot drop and flexor plantar responses

➢ Labs/Imaging
  - Hemoglobin 11.9. Otherwise, complete blood count, basic chemistry panel, liver function tests are unremarkable.
  - Chest x-ray shows borderline cardiomegaly.
  - ECG, NSR, 1st degree AVB, LBBB

HOSPITAL COURSE

➢ In the post-anesthesia care unit, she has paroxysms of hypotension and bradycardia that progresses to complete heart block.
➢ These complications resolve with supportive measures, including atropine, lidocaine, and electrolyte repletion.
➢ She subsequently develops apnea and hypoxic respiratory failure, requiring mechanical ventilation.
➢ After several days, the patient is unable to wean from the ventilator.
➢ Myasthenia gravis is considered, and the patient is started on pyridostigmine without improvement; acetylcholine antibodies are negative.
➢ Brain magnetic resonance imaging is unremarkable.
➢ An electromyogram shows myotonia and sensorimotor axonal neuropathy.
➢ Muscle biopsy is consistent with myotonic dystrophy and lipid myopathy.
➢ Genetic testing confirms the patient has type one myotonic dystrophy.
➢ The patient requires a tracheostomy, and weaning from ventilation takes almost two months.
➢ Supportive care is implemented for four weeks, and the patient has a pulseless electrical activity code with anoxic brain injury.
➢ The patient died two months after admission. Her body was donated to the Myotonic Dystrophy Foundation.

DISCUSSION

➢ Myotonic dystrophy is an autosomal dominant condition with a prevalence of 1:8000. It results from an expanded CTG trinucleotide repeat located in an untranslated portion of the dystrophia myotonica protein kinase (DMPK) gene and demonstrates genetic anticipation.
➢ Several proteins are altered, including the skeletal muscle chloride channel, insulin receptor, and cardiac troponin T.
➢ Our patient presented with classic features including facial and distal muscle weakness, myotonia (seen best in the hands), cataracts, insulin resistance, cardiac conduction and structural abnormalities, excessive somnolence and respiratory failure (sometimes precipitated by general anesthesia).
➢ Patients often present to internists and other subspecialties before seeing neurologists. Treatment is supportive, with no disease modifying therapy available.
➢ Always consider neurological causes of respiratory failure when other more common causes do not seem likely.

REFERENCES AVAILABLE UPON REQUEST