A case of recurrent and progressive respiratory failure

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Introduction: Patients presenting with dyspnea are common. Often times, patients carry previously anchored diagnoses, such as COPD, as a cause of their pulmonary symptoms. It is important, however, to perform a thorough history and physical examination in order to consider less common causes of dyspnea, such as in the case of this patient who was diagnosed with amyotrophic lateral sclerosis (ALS). The typical course for this disease process includes progressive limb and bulbar muscular weakness with eventual involvement of the respiratory musculature, ultimately leading to respiratory failure - the most frequent cause of death in ALS within 2 to 5 years of diagnosis.

Case description: A 68-year-old woman presented to the hospital with recurrent episodes of dyspnea and carbon dioxide retention. She has a history of type II diabetes, hyperlipidemia, hypertension, cervical stenosis, and chronic obstructive pulmonary disease (COPD) requiring home oxygen therapy. Her medications included the following COPD regimen: short acting anticholinergic/beta agonist inhaler, mucolytic, steroid, long acting beta agonist nebulizers, and a Trilogy adaptive servo-ventilation device for nighttime breathing assistance. In the ED, workup showed pH 7.34, pCO2 95mmHg, and HCO3 of 50mmHg; chest x-ray was significant with mild hyperinflation of the lungs. On examination, she had mild proximal upper extremity weakness, bilateral tachypnea, and a nasal voice. She was in the ICU for 24-48 hours for intensive positive pressure therapy because of her severe carbon dioxide retention.

Results and conclusions: Bedside pulmonary function testing was consistent with a restrictive process, and she was diagnosed with obesity hypoventilation. However, her BMI was only 39, and given her history of weakness and tachypnea, we were concerned for a neurologic process. Neurology found fibrillations with insertion and prominent fasciculations within the proximal right upper limb muscles on needle electromyography. Ultrasound examination with phrenic nerve stimulation showed reduced recruitment of large, complex motor unit potentials in both hemidiaphragm and intercostal muscles. With this constellation of symptoms, ALS was diagnosed. Other possible diagnoses were ruled out with neuroimaging, serologic, and cerebrospinal fluid studies.

Take-home message: Progressive dyspnea as the major presenting symptom of ALS is exceedingly rare, occurring in less than 1% according to literature. It is important to keep ALS in the differential diagnosis in patients who present with progressive dyspnea and restrictive lung disease on pulmonary function testing because this diagnosis has significant prognostic difference compared to other entities such as obesity hypoventilation syndrome.

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Macrophage activation syndrome in a Case of dermatomyositis overlapping syndrome with systemic lupus erythematosus: A case report

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Introduction: Macrophage activation syndrome (MAS) is a rare but aggressive life-threatening auto-immune disease. It is characterized by fever, rash, splenomegaly, blood cytopenia, hypertriglyceridemia, high ferritin levels, liver insufficiency, coagulopathy and neurologic involvement. Persistent activation of inflammatory cells like macrophages, natural killer cells and cytotoxic lymphocytes can lead to a cytokine storm and multi organ damage. MAS is usually triggered by rheumatologic diseases and rarely in the presentation of a new connective disease like systemic lupus erythematosus (SLE). In addition to MAS, the auto-immune conditions of SLE can be associated with different overlapping syndromes notably dermatomyositis.

Case description: We present a 31-year-old male from a Latin-American background without pre-existent conditions who presented complain of sore throat, joint pain, fever and fatigue. He quickly developed a pancytopenia with increased liver and pancreatic enzymes. In the process of the investigation, he was treated with antibiotics and admitted intubated to the intensive care unit for a severe pneumonia.

Result and conclusion: We proceeded with a bone marrow biopsy which detected an active MAS. Regarding his muscle weakness, we also revealed an inflammatory myositis on a quadripel muscle biopsy. Further discovery of positive auto-antibodies (ANA and anti-DNA) showed the presence of a LED. We successfully treated his different auto-immune complications with high doses of prednisone, and intravenously immunoglobulins. After 2 months of his admission and 12 days passed in the intensive care unit, the patient returned home with minimal sequelae with a long term immunosuppressive treatment of prednisone, mycophenolate mofetil and hydroxychloroquine.

Take-home message: The early identification of the cause of MAS is crucial for the accurate management of this disease and preventing further multi organ complications. SLE has remains a complex condition that can present its first manifestations in a broad spectrum of auto-immune diseases.

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Gastrointestinal metastases from breast cancer: A case report

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Background: Breast cancer is the most common type of cancer in women nowadays. According to published major studies, the most common sites of metastases of breast cancer are bone, lung, liver and brain. However, it can also metastasize rarely to the gastrointestinal tract. Among the different subtypes of breast cancer, gastrointestinal spread has been associated to infiltrating lobular carcinoma. We present a case of perforated acute diverticulitis that underwent surgery, in which the pathological exam informed of colonic metastasis of lobular breast carcinoma.

Case report: A 78-year-old woman, with medical history of high blood pressure, diabetes and left mastectomy performed 14 years ago for infiltrating lobular carcinoma (Stage T2N2MO), with positive estrogen receptors. Oncological controls showed pleural and bone progression in the last year, so hormonal therapy was indicated. She was admitted to the emergency department due to 72 hours of left lower-quadrant abdominal pain associated with constipation and nausea. On examination she presented tenderness and a palpable mass in the left lower quadrant. Blood tests showed an increased leukocyte count of 13.5x103/μL with neutrophilia, a CRP of 356mg/L and high lactate levels (4.5mmol/L). An abdominal computed tomography (CT) scan showed a left inguinal abscess (6x8x7cm) communicating with an inflammatory mass involving the sigmoid colon, as well as extensive bone metastases, not visualized in previous CTs. An emergency Hartmann’s procedure was performed. The post-operative period was uneventful. The pathological report of the surgical specimen informed of infiltration in multiple diverticula by a carcinoma, with morphological pattern and immunohistochemistry compatible with a lobular breast carcinoma. The patient was derived to the Department of Oncology to continue follow-up and hormonal therapy.