

DSS Study
Correct Diagnoses with
Gold Standards/Excluded Findings and
Initial Rated Difficulty

Case 001 Diagnosis: Acromegaly (diabetic ketoacidosis)

Rated Difficulty: 6.00

Removed Findings: Obvious diagnosis = diabetic ketoacidosis. But, additional labs showed: random growth hormone level twice the upper limit of normal; growth hormone level abnormally elevated after suppression with oral glucose; lateral films of the feet with increased heel pad thickness; and CT of the head with a sella turcica mass without suprasellar extension. Final diagnosis: acromegaly.

Case 011 Diagnosis: Blastomycosis

Rated Difficulty: 5.83

Removed Findings: Skin swabs and biopsies of the leg ulcer showed blastomycosis. He was treated with Amphotericin B, with improvement. Diagnosis: Blastomycosis.

Case 012 Diagnosis: Metastatic Hepatic Adeno (liver) Cancer

Rated Difficulty: 5.00

Removed Findings: A CT scan that was almost definitive, available 2-3 days into hospitalization, and a liver biopsy.

Case 021 Diagnosis: Carcinoid Syndrome

Rated Difficulty: 4.33

Removed Findings: A 24-hour urine for 5-hydroxyindoleacetic acid (5HIAA) was 146 (normal, 0 to 10). An abdominal ultrasound revealed hepatomegaly with multiple echogenic masses and dilated intrahepatic ducts. An abdominal CT scan showed a questionable pancreatic mass. A lower GI revealed a mass in the terminal ileum. Colonoscopy revealed a 2.5 cm mass with a broad base at the cecum; a biopsy revealed carcinoid. A liver biopsy also showed carcinoid. Diagnosis: carcinoid syndrome.

Case 022 Diagnosis: Non-Hodgkins Lymphoma

Rated Difficulty: 4.67

Removed Findings: (1) A repeat abdominal CT scan that was obtained shortly after admission. It revealed a renal pelvis mass on the left and some increase in the lymphadenopathy and splenomegaly that had been noted on the earlier scan. (2) A bone marrow biopsy which showed hypercellularity in all lines and no malignant cells.

Case 023 Diagnosis: Hairy Cell Leukemia

Rated Difficulty: 6.00

Removed Findings: Bone marrow aspirate and biopsy. Diagnosis: Hairy Cell Leukemia.

Case 031 Diagnosis: Cryoglobulinemia

Rated Difficulty: 6.00

Removed Findings: Additional workup showed that serum was positive for cryoglobulins, with a cryocrit of 20% (*cold panel cryocrit is 20%*). A renal biopsy revealed crescent formation and increased mesangial matrix. Stains for amyloid were negative. A bone marrow biopsy was normal.

Diagnosis: Cryoglobulinemia, with crescentic glomerulonephritis.

Case 032 Diagnosis: Ulcerative Colitis

Rated Difficulty: 3.50

Removed Findings: A colonoscopy.

Case 033 Diagnosis: Thrombotic Thrombocytopenic Purpura

Rated Difficulty: 3.67

Removed Findings: Contained within the case scenario: Intravascular Hemolysis: Mechanical. Diagnosis: Thrombotic Thrombocytopenic Purpura (TTP)

Case 042 Diagnosis: Appendicitis

Rated Difficulty: 3.67

Removed Findings: A laparoscopy that revealed appendicitis

Case 043 Diagnosis: Amoebic Liver Abscess

Rated Difficulty: 3.67

Removed Findings: Amebic serologies Diagnosis: Amoebic liver abscess.

Case 052 Diagnosis: Colon Cancer

Rated Difficulty: 3.17

Removed Findings: Laboratory data that showed iron-deficiency anemia and a colonoscopy/biopsy that showed malignancy.

Case 053 Diagnosis: Crohn's Disease

Rated Difficulty: 4.17

Removed Findings: Laparotomy with biopsy. Diagnosis: Crohn's disease.

Case 062 Diagnosis: Cryptococcal Meningitis

Rated Difficulty: 3.83

Removed Findings: A lumbar puncture/CSF examination under india ink that revealed cryptococcus.

Case 063 Diagnosis: Brucellosis

Rated Difficulty: 5.67

Removed Findings: Brucella serology. Diagnosis: Brucellosis.

Case 072 Diagnosis: Hemolytic Uremic Syndrome

Rated Difficulty: 5.50

Removed Findings: Renal biopsy that showed thrombotic microangiopathy.

Case 082 Diagnosis: Miliary (disseminated) TB

Rated Difficulty: 5.67

Removed Findings: Non-pulmonary culture that was positive for TB.

Case 083 Diagnosis: Aortic Dissection

Rated Difficulty: 5.17

Removed Findings: CT scan. Diagnosis: Aortic dissection.

Case 091 Diagnosis: Guillain-Barre Syndrome

Rated Difficulty: 3.33

Removed Findings: An edrophonium (Tensilon) test was negative; antiacetylcholine receptor antibody was not detected. EMG/NCV showed decreased nerve conduction velocities. Nerve and muscle biopsy showed focal loss of myelin and atrophy of type II muscle fibers; there was no evidence of vasculitis. Diagnosis: Guillain-Barre syndrome.

Case 092 Diagnosis: Osteomalacia

Rated Difficulty: 5.50

Removed Findings: Vitamin D deficiency based on serum vitamin D levels and a bone biopsy that revealed osteomalacia.

Case 093 Diagnosis: Subarachnoid Hemorrhage

Rated Difficulty: 4.50

Removed Findings: Lumbar puncture. Diagnosis: subarachnoid hemorrhage.

Case 102 Diagnosis: Pheochromocytoma

Rated Difficulty: 4.00

Removed Findings: A CT scan of adrenals and an adrenal biopsy that were positive for pheochromocytoma.

Case 103 Diagnosis: Cardiac Amyloidosis

Rated Difficulty: 5.67

Removed Findings: Endocardial biopsy. Diagnosis: Cardiac amyloidosis.

Case 111 Diagnosis: Mucormycosis

Rated Difficulty: 4.00

Removed Findings: A KOH prep of scrapings from the hard palate revealed hyphae (*biopsy of suspected tissue is positive for zygomycetes*). Palatal biopsies showed hyphae invading the tissue; the appearance was consistent with Mucor. Diagnosis: Mucormycosis.

Case 112 Diagnosis: Whipple's Disease

Rated Difficulty: 6.17

Removed Findings: A small bowel biopsy that showed PAS positive for macrophages.

Case 113 Diagnosis: Hemachromatosis

Rated Difficulty: 3.67

Removed Findings: Liver biopsy. Diagnosis: hemachromatosis.

Case 121 Diagnosis: Myasthenia Gravis

Rated Difficulty: 3.17

Removed Findings: An edrophonium (Tensilon) test revealed dramatic improvement in his ptosis. Electromyography showed decremental changes in muscle action potentials. Acetyl choline-receptor antibodies were negative. A CT scan of the chest showed an anterior mediastinal mass, consistent with a thymoma. At surgery, a large thymoma that was adherent to lung and pericardial tissue was resected. Pathology revealed a benign thymoma with reactive lymphoid hyperplasia. He was discharged on pyridostigmine and prednisone. Diagnosis: Myasthenia gravis.

Case 122 Diagnosis: Hypokalemic Periodic Paralysis

Rated Difficulty: 4.67

Removed Findings: Increased serum potassium (24 hour urine for potassium).

Case 123 Diagnosis: Syphilitic Meningitis

Rated Difficulty: 6.00

Removed Findings: VDRL/RPR. Diagnosis: syphilitic meningitis.

Case 133 Diagnosis: Pernicious Anemia

Rated Difficulty: 3.67

Removed Findings: Serum B12 level. Diagnosis: Pernicious anemia.

Case 143 Diagnosis: Polymyalgia Rheumatica

Rated Difficulty: 4.00

Removed Findings: Trial of low dose steroids. Diagnosis: Polymyalgia rheumatica.

Case 161 Diagnosis: Porphyria (cutanea tarda)

Rated Difficulty: 4.17

Removed Findings: Urinary δ -aminolevulinic acid (ALA) was 4.6 (normal, 1.5 to 7.5); porphobilinogen was negative; coproporphyrin was 220 (normal, 0 to 180), and uroporphyrin was 2,123 (normal, 0 to 60). Stool coproporphyrin was 96, and protoporphyrin was 5.3. Diagnosis: Porphyria cutanea tarda.

Case 171 Diagnosis: Silicosis

Rated Difficulty: 3.67

Removed Findings: Bronchoscopy and biopsy showed changes compatible with silicosis. Pleural biopsy showed chronic inflammation with neutrophilic and lymphocytic infiltration, and some granuloma-like structures; there were no acid-fast bacilli. Final diagnosis: silicosis.

Case 181 Diagnosis: Temporal Arteritis

Rated Difficulty: 5.17

Removed Findings: Temporal artery biopsy revealed granulomas consistent with temporal arteritis. The patient was started on prednisone, with resolution of his fever, improvement of his symptoms, and a decrease in the sedimentation rate. Diagnosis: Temporal arteritis.

Case 251 Diagnosis: Amyloidosis (renal)

Rated Difficulty: 4.83

Removed Findings: Renal biopsy, gingival biopsy, and biopsy of the tongue lesion revealed amyloidosis.

Case 291 Diagnosis: Gaucher's Disease

Rated Difficulty: 6.17

Removed Findings: A bone marrow examination showed a hypercellular marrow with severe erythroid hyperplasia. Gaucher's cells were present. Nontartrate acid phosphatase was 0.4. Serum glucocerebrosidase was 0.01 (normal, 0.06 to 0.1). Diagnosis: Gaucher's disease.

Notes:

- Cases are from three medical centers
- Removed findings indicates the findings that were removed to make the case more challenging.
- Rated difficulty: Average difficulty rating of three clinicians on a seven point scale (1-7). These differ somewhat from actual empirical difficulty.