

Case 13 (April 27 2006): [N Engl J Med 2006;354:1828-37.](#)

Case 13-2006 — A 50-Year-Old Man with a Painful Bone Mass and Lesions in the Liver

Demographic: 50 year old, male, America North, General speciality

Clinical features extracted from the case and entered into the query box:

4 months previously:

"elbow pain"
Stiffness in elbow
progressive pancytopenia

2 months previously:

"elbow pain"
Stiffness in elbow
progressive pancytopenia
radiographs showed lytic lesion in proximal ulna
"proximal ulna" tender and posterior soft- tissue mass
decreased range of movement in elbow
palpable liver in right upper quadrant
"ulna fracture"
CT of chest, abdomen and pelvis showed multiple masses

Admission:

pain and stiffness in the elbow exacerbated by movement
tender "proximal ulna"
hepatomegaly
decreased "range of movement" in elbow
lytic lesion in the "proximal ulna" with pathologic fracture
ulna cortical destruction
soft-tissue mass below elbow
multiple hepatic masses
"hypocellular marrow"
elevated alphafetoprotein
progressive pancytopenia

Synonyms used by Isabel (optional) for the above query:

Admission:

Range of movement
Hepatomegaly

STOP !

Before you read further you might want to construct your own:

- Complete differential diagnosis
- Final diagnosis

In the following section you will find:

- The differential diagnosis constructed by the MGH panel
- The final diagnosis of the case
- Which of the MGH panel differential diagnoses did Isabel contain
- Did Isabel suggest the Final diagnosis

Differential Diagnoses considered by the MGH panel:

Malignant bone tumour
Hepatic Cancer – hepatocellular cancer
Fanconi's Anemia

Final Diagnosis of the case according to NEJM:

Fanconi's Anemia and Hepatocellular Carcinoma metastatic to the bone.

Differential Diagnoses of the case as given by Isabel:

Malignant bone tumour – metastasis – Isabel 1st page
Hepatocellular failure – Isabel 2nd page

Was the final diagnosis given by Isabel?

Yes, Malignant Bone Tumours - Bone Metastases (Isabel 1st page)

Entire presentation cut and pasted into the Isabel query box:

None of the above

Isabel Differential:

<p>ORTHOPEDIC DISORDERS</p> <p>Benign Bone Tumors <input type="checkbox"/></p> <p>Vit D Deficient Rickets/Osteomalacia <input type="checkbox"/></p> <p>Renal Osteodystrophy <input type="checkbox"/></p> <p>↳ Malignant Bone Tumours <input type="checkbox"/></p> <p>↳ Bone Osteochondroma <input type="checkbox"/></p> <p>Hypophosphatasia <input type="checkbox"/></p> <p>Paget's Disease <input type="checkbox"/></p> <p>Elbow Injuries <input type="checkbox"/></p> <p>Engelmann's Disease <input type="checkbox"/></p> <p>Osteopetrosis <input type="checkbox"/></p> <p>Frozen Shoulder <input type="checkbox"/></p>	<p>NERVOUS SYSTEM DISORDERS</p> <p>Ulnar Nerve Injury <input type="checkbox"/></p> <p>Intracranial Hypertension <input type="checkbox"/></p>
	<p>NEOPLASTIC DISEASES</p> <p>Langerhans Cell Histiocytosis Class I <input type="checkbox"/></p>
	<p>RESPIRATORY SYSTEM DISORDERS</p> <p>↳ Lung Neoplasms <input type="checkbox"/></p> <p>Sarcoidosis <input type="checkbox"/></p>
	<p>NEUROMUSCULAR DISORDERS</p> <p>Dermatomyositis / Polymyositis <input type="checkbox"/></p>
<p>HUMAN GENETICS</p> <p>↳ Heart-Hand Syndrome <input type="checkbox"/></p>	<p>RHEUMATIC DISEASES</p> <p>↳ Osteomyelitis and Septic Arthritis <input type="checkbox"/></p> <p>Mixed Connective Tissue Disease <input type="checkbox"/></p> <p>Pyogenic Arthritis <input type="checkbox"/></p> <p>↳ Spondyloarthropathies <input type="checkbox"/></p> <p>↳ Systemic Sclerosis <input type="checkbox"/></p> <p>Behçet's Syndrome <input type="checkbox"/></p> <p>Rheumatoid Arthritis <input type="checkbox"/></p>
<p>UROLOGIC DISORDERS</p> <p>↳ Neoplasms of the Kidney <input type="checkbox"/></p>	
<p>DERMATOLOGY</p> <p>Psoriasis <input type="checkbox"/></p>	
<p>INFECTIOUS DISEASES</p> <p>Gonococcal Infection <input type="checkbox"/></p>	
<p>METABOLIC DISEASES</p> <p>↳ Primary Dyslipidemias <input type="checkbox"/></p>	<p>GASTROINTESTINAL DISORDERS</p> <p>Hepstocellular Failure <input type="checkbox"/></p>