

NEJM 14 (May 11 2006): [N Engl J Med 2006;354:2047-56.](#)

Case 14-2006 — A 25-Year-Old Woman with Anemia and Iron Overload

Demographic: 25 yrs, female, not pregnant, America North,
General speciality

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

normochromic, normocytic anemia
iron overload
low reticulocyte count
high ESR

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

high ESR
iron overload

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:

Diamond–Blackfan anemia
 β -Thalassemia

Final Diagnosis of the case according to NEJM:

$\delta\beta$ -thalassemia and Diamond–Blackfan anemia

Differential Diagnoses of the case as given by Isabel:

Diamond–Blackfan Anemia (Congenital Hypoplastic Anemia) –
Isabel 1st page
Thalassemia syndromes– Isabel 1st page

Was the final diagnosis given by Isabel?

Yes, DBA (Isabel 1st page).

Entire presentation cut and pasted into the Isabel query box:

Diamond–Blackfan Anemia (Congenital Hypoplastic Anemia) –
Isabel 1st page
Beta-Thalassemia – Isabel 1st page

Isabel Differential:

HEMATOLOGY	INFECTIOUS DISEASES
Anemia of Chronic Disorders <input type="checkbox"/>	EBV Infection <input type="checkbox"/>
Congenital Hypoplastic Anemia <input type="checkbox"/>	Q Fever <input type="checkbox"/>
Hereditary Elliptocytosis <input type="checkbox"/>	Babesiosis <input type="checkbox"/>
Thalassemia Syndromes <input type="checkbox"/>	↳CSF Shunt Disorders <input type="checkbox"/>
PNH <input type="checkbox"/>	
G6PD Deficiency <input type="checkbox"/>	RESPIRATORY SYSTEM DISORDERS
Pyruvate Kinase Deficiency <input type="checkbox"/>	Lung Abscess <input type="checkbox"/>
NEPHROLOGY	UROLOGIC DISORDERS
Glomerulonephritis <input type="checkbox"/>	↳Upper Urinary Tract Obstruction <input type="checkbox"/>
GASTROINTESTINAL DISORDERS	RHEUMATIC DISEASES
Hepatocellular Failure <input type="checkbox"/>	Sjögren's Syndrome <input type="checkbox"/>
	SLE <input type="checkbox"/>
NEOPLASTIC DISEASES	GYNECOLOGICAL DISORDERS
↳Pancreatic Neoplasms <input type="checkbox"/>	Pelvic Inflammatory Disease <input type="checkbox"/>
↳Leukemia <input type="checkbox"/>	