

**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  
 $\beta$ -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 1<sup>st</sup> page  
Thalassemia syndromes– Isabel 1<sup>st</sup> page

***Was the final diagnosis given by Isabel?***

Yes, DBA (Isabel 1<sup>st</sup> page).

***Entire presentation cut and pasted into the Isabel query box:***

Diamond–Blackfan Anemia (Congenital Hypoplastic Anemia) –  
Isabel 1<sup>st</sup> page  
Beta-Thalassemia – Isabel 1<sup>st</sup> page

***Isabel Differential:***

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