

**Case 33 (October 26 2006):** [N Engl J Med 2006;355:1812-9](#)

**Demographic:** Male, 43yrs, America North, General speciality

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

hypogonadism  
gradual onset of fatigue  
decreased libido  
erectile dysfunction  
dry mouth  
polyuria  
hyperpigmentation  
arthralgia in ankles  
weight loss  
thrombocytopenia  
hyperglycemia  
high ALT  
high AST  
high ALP  
high serum iron  
low iron binding capacity  
high ferritin  
low Hb

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

decreased libido  
thrombocytopenia  
erectile dysfunction  
weight loss  
low hb  
hyperglycemia

**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:***

Primary (hereditary) hemochromatosis  
Secondary hemochromatosis

***Final Diagnoses of the case according to NEJM:***

Hereditary hemochromatosis due to a homozygous C282Y mutation in the *HFE* gene, with hepatic cirrhosis, diabetes, and hypogonadism.

***Differential Diagnoses of the case as given by Isabel:***

Hemochromatosis – Isabel 1<sup>st</sup> page  
Diabetes Type 2 – Isabel 1<sup>st</sup> page

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

Yes, Isabel 1<sup>st</sup> page

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

Diabetes Type 2 – Isabel 1<sup>st</sup> page

***Isabel differential for extracted features:***

<p><b>HEMATOLOGY</b> &lt;&lt; <a href="#">Click here</a></p> <ul style="list-style-type: none"> <li>↳ <a href="#">Hemochromatosis</a> <span style="float: right;">RD</span></li> <li>↳ <a href="#">Sideroblastic Anemias</a> <span style="float: right;">RD</span></li> <li><a href="#">Anemia of Chronic Disorders</a> <span style="float: right;">RD</span></li> </ul>	<p><b>INFECTIOUS DISEASES</b> &lt;&lt; <a href="#">Click here</a></p> <ul style="list-style-type: none"> <li><a href="#">Brucellosis</a> <span style="float: right;">RD</span></li> <li><a href="#">Vibrio Vulnificus Infection</a> <span style="float: right;">RD</span></li> <li>↳ <a href="#">CMV Infections</a> <span style="float: right;">RD</span></li> </ul>
<p><b>RESPIRATORY SYSTEM DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p> <ul style="list-style-type: none"> <li>↳ <a href="#">Sleep Apnea</a> <span style="float: right;">RD</span></li> <li><a href="#">Sarcoidosis</a> <span style="float: right;">RD</span></li> </ul>	<p><b>ENDOCRINE SYSTEM</b> &lt;&lt; <a href="#">Click here</a></p> <ul style="list-style-type: none"> <li>↳ <a href="#">Sexual Dysfunction</a> <span style="float: right;">RD</span></li> <li><a href="#">Type 2 Diabetes</a> <span style="float: right;">RD</span></li> </ul>
<p><b>LIVER DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>RHEUMATIC DISEASES</b> &lt;&lt; <a href="#">Click here</a></p>
<p><b>DERMATOLOGY</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>GASTROINTESTINAL DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p>
<p><b>TOXICOLOGY</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>NUTRITIONAL DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p>
<p><b>NEPHROLOGY</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>ALLERGIC DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p>

**Isabel differential for features as summarised Dr. Raymond T. Chung:**

<b>HEMATOLOGY</b> << <a href="#">Click here</a> ↳ <u>Hemochromatosis</u> 45% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Sideroblastic Anemias</u> 27% <input type="checkbox"/> <input checked="" type="checkbox"/> RD	<b>INFECTIOUS DISEASES</b> << <a href="#">Click here</a> ↳ <u>Vibrio Vulnificus Infection</u> 35% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Viral Hepatitis</u> 30% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Meningococcal Disease</u> 28% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Lyme Disease</u> 25% <input type="checkbox"/> <input checked="" type="checkbox"/> RD
<b>ENDOCRINE SYSTEM</b> << <a href="#">Click here</a> ↳ <u>Primary Adrenal Insufficiency</u> 32% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Gonadotropin Secreting Tumors</u> 25% <input type="checkbox"/> <input checked="" type="checkbox"/> RD ↳ <u>Sexual Dysfunction</u> 25% <input type="checkbox"/> <input checked="" type="checkbox"/> RD	<b>LIVER DISORDERS</b> << <a href="#">Click here</a>
<b>NEOPLASTIC DISEASES</b> << <a href="#">Click here</a> ↳ <u>Leukemia</u> 25% <input type="checkbox"/> <input checked="" type="checkbox"/> RD	<b>CARDIAC DISORDERS</b> << <a href="#">Click here</a>
<b>GASTROINTESTINAL DISORDERS</b> << <a href="#">Click here</a>	<b>HUMAN GENETICS</b> << <a href="#">Click here</a>
<b>RESPIRATORY SYSTEM DISORDERS</b> << <a href="#">Click here</a>	<b>TOXICOLOGY</b> << <a href="#">Click here</a>
<b>NERVOUS SYSTEM DISORDERS</b> << <a href="#">Click here</a>	<b>METABOLIC DISEASES</b> << <a href="#">Click here</a>
<b>RHEUMATIC DISEASES</b> << <a href="#">Click here</a>	

***Isabel differential for classic features of Hemochromatosis:***

<p><b>HEMATOLOGY</b> &lt;&lt; <a href="#">Click here</a></p> <p>↳ <u>Hemochromatosis</u> 62% <input type="checkbox"/> <b>RD</b></p>	<p><b>CARDIAC DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p> <p>↳ <u>Myocarditis</u> 28% <input type="checkbox"/> <b>RD</b></p> <p>↳ <u>Infective Endocarditis</u> 28% <input type="checkbox"/> <b>RD</b></p> <p>↳ <u>Pericarditis</u> 25% <input type="checkbox"/> <b>RD</b></p> <p>↳ <u>Cardiomyopathy</u> 24% <input type="checkbox"/> <b>RD</b></p>
<p><b>NEOPLASTIC DISEASES</b> &lt;&lt; <a href="#">Click here</a></p> <p>↳ <u>Non-Hodgkin's Lymphoma</u> 28% <input type="checkbox"/> <b>RD</b></p>	
<p><b>LIVER DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p> <p>↳ <u>Cirrhosis</u> 25% <input type="checkbox"/> <b>RD</b></p>	<p><b>RESPIRATORY SYSTEM DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p> <p><u>Sarcoidosis</u> 27% <input type="checkbox"/> <b>RD</b></p>
<p><b>RHEUMATIC DISEASES</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>INFECTIOUS DISEASES</b> &lt;&lt; <a href="#">Click here</a></p> <p><u>Lyme Disease</u> 25% <input type="checkbox"/> <b>RD</b></p> <p><u>Babesiosis</u> 25% <input type="checkbox"/> <b>RD</b></p>
<p><b>TOXICOLOGY</b> &lt;&lt; <a href="#">Click here</a></p>	
<p><b>DERMATOLOGY</b> &lt;&lt; <a href="#">Click here</a></p>	<p><b>ENDOCRINE SYSTEM</b> &lt;&lt; <a href="#">Click here</a></p>
	<p><b>HUMAN GENETICS</b> &lt;&lt; <a href="#">Click here</a></p>
	<p><b>ALLERGIC DISORDERS</b> &lt;&lt; <a href="#">Click here</a></p>