

**Case 23 (July 27 2006):** [N Engl J Med 2006;355:394-402](#)

**Demographic:** Male, 36 yrs, America North, General speciality

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

numbness in arm  
sustained hypertension  
profuse sweating at night  
palpitations  
insomnia  
Anxiety  
retinal hemangioma in the superotemporal periphery, with a large vein  
adrenal mass  
complex cyst in kidney with enhancing septations.  
multiple hemangioma lesions in liver  
medullary tumor  
small focal area of enhancement posterior to the spinal cord

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

insomnia

**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:***

Recurrent pheochromocytoma (hereditary)  
Multiple Endocrine Neoplasia  
Neurofibromatosis  
Familial Paraganglioma  
von Hippel–Lindau Disease (VHL)

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

VHL disease with pheochromocytoma, hemangioblastomas of the medulla and spinal cord, and renal and hepatic cysts.

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

Recurrent pheochromocytoma – Isabel 1<sup>st</sup> page  
Multiple Endocrine Neoplasia – Isabel 1<sup>st</sup> page  
Neurofibromatosis – Isabel 1<sup>st</sup> page  
Von Hippel–Lindau Disease (VHL) – Isabel 2<sup>nd</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:***

Multiple Endocrine Neoplasia – Isabel 1<sup>st</sup> page

**Isabel differential:**

<b>NERVOUS SYSTEM DISORDERS</b> ∨		
↳ <a href="#">Cervical Spondylosis</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Syringomyelia</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">CVA / Stroke</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Spinal Cord Tumors</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Carpal Tunnel Syndrome</a>	<input type="checkbox"/>	<input type="checkbox"/> R
↳ <a href="#">Ataxic Disorders</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Multiple Sclerosis</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Progressive Supranuclear Palsy</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Ekbom's Syndrome</a>	<input type="checkbox"/>	<input type="checkbox"/> R
↳ <a href="#">Myelitis</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Intracranial Hemorrhage</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Diabetic Neuropathy</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Thoracic Outlet Syndrome</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>RHEUMATIC DISEASES</b> ∨		
<a href="#">Fibromyalgia</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>NUTRITIONAL DISORDERS</b> ∨		
<a href="#">Thiamine Deficiency</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>INFECTIOUS DISEASES</b> ∨		
<a href="#">Lyme Disease</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>VASCULAR DISORDERS</b> ∨		
<a href="#">Peripheral Arterial Disease</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>NEUROMUSCULAR DISORDERS</b> ∨		
↳ <a href="#">Polyneuropathy Disorders</a>	<input type="checkbox"/>	<input type="checkbox"/> R
↳ <a href="#">Hereditary Motor-Sensory Neuropathies</a>	<input type="checkbox"/>	<input type="checkbox"/> R
↳ <a href="#">Motor Neurone Disease</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Dermatomyositis / Polymyositis</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Endocrine Myopathies</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Familial Spastic Paraplegia</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>RESPIRATORY SYSTEM DISORDERS</b> ∨		
↳ <a href="#">Lung Neoplasms</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Sarcoidosis</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>ENDOCRINE SYSTEM</b> ∨		
<a href="#">Hypothyroidism</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>ORTHOPEDIC DISORDERS</b> ∨		
↳ <a href="#">Degenerative Spinal Disorders</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<a href="#">Intervertebral Disk Herniation</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>HEMATOLOGY</b> ∨		
↳ <a href="#">Megaloblastic Anemias</a>	<input type="checkbox"/>	<input type="checkbox"/> R
<b>IMMUNOLOGIC SYSTEM</b> ∨		
<a href="#">Eosinophilia-Myalgia Syndrome</a>	<input type="checkbox"/>	<input type="checkbox"/> R