

Case 18 (June 15 2006): [N Engl J Med 2006;354:2584-92](#)

Demographic: Female, 57 yrs, Not-pregnant, America North, General Speciality

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

chronic progressive numbness and weakness in feet and legs
progressive gait difficulty
orthostatic hypotension
hammer toes
head tremor
unable to walk on toes or heels
Romberg sign present
bilaterally reduced strength in the dorsiflexor, invertor, and evertor muscles in feet
deep-tendon reflexes absent
pupils dilated and unreactive to light or accommodation
reduced sensation to light touch in feet up to proximal legs
reduced sensation to pinprick to middle of legs
position sense and vibration sensation absent in toes and reduced at ankles

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

Orthostatic hypotension

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:

Polyneuropathy

- Diabetes / Impaired Glucose intolerance
- Acute polyneuropathy - variant of the Guillain-Barré syndrome.
- Subacute polyneuropathy - suggest a toxic or perhaps a metabolic cause for the neuropathy, such as exposure to heavy metals or vitamin B12 deficiency.
- Chronic polyneuropathy – associated with diabetes or monoclonal gammopathy but is more likely to be inherited or idiopathic. Charcot-Marie- Tooth disease

Final Diagnoses of the case according to NEJM:

Charcot-Marie- Tooth disease Type 2

Differential Diagnoses of the case as given by Isabel:

Polyneuropathy – Isabel 1st page

HMSN – Charcot-Marie Tooth disease– Isabel 1st page

Diabetic neuropathy – Isabel 1st page

Vitamin B12 deficiency (Megaloblastic Anemias)– Isabel 1st page

Was the final diagnosis given by Isabel:

Yes, Isabel 1st page

Entire presentation cut and pasted into the Isabel query box:

Polyneuropathy – Isabel 1st page

HMSN – Charcot-Marie Tooth disease– Isabel 1st page

Diabetic neuropathy – Isabel 1st page

Isabel differential:

<p>NEUROMUSCULAR DISORDERS</p> <ul style="list-style-type: none"> ↳ Polyneuropathy Disorders <input type="checkbox"/> ↳ Hereditary Motor-Sensory Neuropathies <input type="checkbox"/> Muscular Dystrophies <input type="checkbox"/> 	<p>NERVOUS SYSTEM DISORDERS</p> <ul style="list-style-type: none"> Diabetic Neuropathy <input type="checkbox"/> ↳ Cervical Spondylosis <input type="checkbox"/> Spinal Cord Tumors <input type="checkbox"/> Multiple Sclerosis <input type="checkbox"/> Syringomyelia <input type="checkbox"/> Sciatic Nerve Injuries <input type="checkbox"/> ↳ Myelitis <input type="checkbox"/> ↳ Intracranial Hemorrhage <input type="checkbox"/> CVA/Stroke <input type="checkbox"/> Paraneoplastic Neuro Synd <input type="checkbox"/> Common Peroneal Nerve Injury <input type="checkbox"/>
<p>NUTRITIONAL DISORDERS</p> <ul style="list-style-type: none"> Thiamine Deficiency <input type="checkbox"/> 	
<p>HEMATOLOGY</p> <ul style="list-style-type: none"> ↳ Megaloblastic Anemias <input type="checkbox"/> 	
<p>PSYCHOLOGIC DISORDERS</p> <ul style="list-style-type: none"> ↳ Psychosomatic Illness <input type="checkbox"/> 	
<p>CARDIOVASCULAR SYSTEM</p> <ul style="list-style-type: none"> Peripheral Arterial Disease <input type="checkbox"/> 	<p>ENDOCRINE SYSTEM</p> <ul style="list-style-type: none"> Hypothyroidism <input type="checkbox"/>
<p>GASTROINTESTINAL DISORDERS</p> <ul style="list-style-type: none"> Hepatocellular Failure <input type="checkbox"/> 	