

Case 36 (November 24 2005): [N Engl J Med 2005;353:2271-80](#)

Demographic: Female, 61 yrs, Not pregnant, America North, General speciality

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

seizure
short stature
hearing loss
lactic acidosis
hypertension
residual aphasia
visual-field loss
altered mental status
dementia
recent strokes
myopathy

Synonyms used by Isabel for the above query:

Hearing loss
Short stature
Hypertension
Visual field loss
Altered mental status

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:

CVA (including lobar intracerebral hemorrhage, cerebral venous sinus thrombosis, and hypertensive encephalopathy)
Mitochondrial encephalomyopathy – MELAS
Status epilepticus
Vascular dementia

Final Diagnoses of the case according to NEJM:

MELAS syndrome

Differential Diagnoses of the case as given by Isabel:

MELAS syndrome (Mitochondrial encephalomyopathy) – Isabel 1st page
CVA / Stroke – Isabel 1st page
Vascular dementia – Isabel 1st page
Intracranial hemorrhage - Isabel 1st page
Status epilepticus - click on Nervous system disorders heading
Cerebral sinus venous thrombosis - click on Nervous system disorders heading

Was the final diagnosis given by Isabel:

Yes, Isabel 1st page

Entire presentation cut and pasted into the Isabel query box:

CVA / Stroke – Isabel 1st page
MELAS syndrome - click on Nervous system disorders heading

Isabel differential:

<p>NERVOUS SYSTEM DISORDERS << Click here</p> <p>↳ <u>Mitochondrial Encephalomyopathies</u> 44% <input type="checkbox"/> <input type="checkbox"/> RD</p> <p>↳ <u>CVA / Stroke</u> 36% <input type="checkbox"/> <input type="checkbox"/> RD</p> <p>↳ <u>Vascular Dementia</u> 27% <input type="checkbox"/> <input type="checkbox"/> RD</p> <p>↳ <u>Transient Ischemic Attack</u> 23% <input type="checkbox"/> <input type="checkbox"/> RD</p> <p>↳ <u>Intracranial Hemorrhage</u> 22% <input type="checkbox"/> <input type="checkbox"/> RD</p>	<p>INFECTIOUS DISEASES << Click here</p> <p>↳ <u>Toxoplasmosis</u> 35% <input type="checkbox"/> <input type="checkbox"/> RD</p>
<p>CARDIAC DISORDERS << Click here</p> <p>↳ <u>Ischemic Heart Disease</u> 21% <input type="checkbox"/> <input type="checkbox"/> RD</p>	<p>ENDOCRINE SYSTEM << Click here</p> <p><u>HHNC / HONK</u> 25% <input type="checkbox"/> <input type="checkbox"/> RD</p> <p>↳ <u>Hypothyroidism</u> 23% <input type="checkbox"/> <input type="checkbox"/> RD</p>
<p>NEPHROLOGY << Click here</p>	<p>ORTHOPEDIC DISORDERS << Click here</p> <p><u>Renal Osteodystrophy</u> 23% <input type="checkbox"/> <input type="checkbox"/> RD</p>
<p>RHEUMATIC DISEASES << Click here</p>	<p>HEMATOLOGY << Click here</p>
<p>TOXICOLOGY << Click here</p>	<p>NEUROMUSCULAR DISORDERS << Click here</p>
	<p>DERMATOLOGY << Click here</p>
	<p>METABOLIC DISEASES << Click here</p>
	<p>TRAUMA INJURIES << Click here</p>

