

Case 12 (April 16 2009): [N Engl J Med. 360\(16\):1656-1665](#)

Demographic: Male, 46 yrs, America North, General speciality

Clinical features extracted from the case and entered into the query

box:

hemiparesis
aphasia
migraines
dysarthria
facial weakness
brisk reflexes
dysphagia
severe fatigue
nausea
vomiting
phonophobia

Synonyms used by Isabel for the above query:

dysphagia
dysarthria
brisk reflexes
fatigue
vomiting
facial weakness
aphasia
hemiparesis
phonophobia

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:

MELAS

Final Diagnosis of the case according to NEJM: Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL).

Differential Diagnoses of the case as given by Isabel:

MELAS –Mitochondrial Encephalomyopathies - under Infectious diseases heading

Was the final diagnosis given by Isabel:

Yes, Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) – under Infectious diseases heading

Isabel Healthcare Inc

3120 Fairview Park Drive, Suite 200, Falls Church, VA 22042

Tel: +1.703.289.8855

Isabel Differential:

- NERVOUS SYSTEM DISORDERS >> click to collapse

- [Migraine](#) ? RD
- [Headache Disorders](#) ? RD
- [CVA / Stroke](#) ? RD
- [Transient Ischemic Attack](#) ? RD
- [Intracranial Abscess](#) ? RD
- [Intracranial Hemorrhage](#) ? RD
- [Head Injury](#) ? RD
- [Multiple Sclerosis](#) ? RD
- [Vascular Dementia](#) ○ RD

Why did this diagnosis come up ? ✕

We matched the terms: dysphagia | dysarthria | slurring of speech | migrain | migraines

Degree of match between query entered and isabel database (Not clinical probability): 33%

...CADASIL

- [Ataxic Disorders](#) ? RD
 - [Cerebral Sinus Venous Thrombosis](#) ? RD
 - [Bell's Palsy](#) ? RD
 - [Mitochondrial Encephalomyopathies](#) ? RD
- #### ...MELAS
- [Cerebral AV Malformations](#) ? RD
 - [HIV Neuro Complications](#) ? RD
 - [Brain Neoplasms](#) ? RD
 - [Aphasic Syndromes](#) ? RD
 - [Progressive Supranuclear Palsy](#) ? RD