

Case 8 (March 17 2011): [N Engl J Med. 364\(11\):1062-1074](#)

Demographic: Female, 32 yrs, North America

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

myoclonic epilepsy
cognitive decline
memory loss
aphasia

Synonyms used by Isabel for the above query:

myoclonic epilepsy
cognitive decline
memory loss
aphasia

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:

Huntingdon's disease

Final Diagnosis of the case according to NEJM:

Kufs' disease (autosomal dominant, Parry type, young-adult-onset neuronal ceroid lipofuscinosis).

Differential Diagnoses of the case as given by Isabel:

Huntingdon's disease under Chorea disorders in Neuro

Was the final diagnosis given by Isabel:

Yes, Neuronal ceroid lipofuscinosis in Neuro

Isabel Checklist Differential:

enter clinical features | synonyms

age*

gender female male

pregnancy

Refine search:

travel

history:

show me:

diagnoses

causative drugs

bioterrorist agents

Enter clinical features, no negatives, no numbers: ⓘ

myoclonic epilepsy
cognitive decline
memory loss
aphasia

+ add a clinical feature

get checklist >

[clear search](#)

Isabel is not meant to replace your clinical judgment.

diagnoses | drugs

Nervous System Disorders

sort by: most relevant | action: -select-

	thumbs up	thumbs down	action
<input checked="" type="checkbox"/> Human Prion Diseases	👍	👎	? NEURO
Frontotemporal Dementia	👍	👎	? NEURO
<input checked="" type="checkbox"/> HIV Neuro Complications	👍	👎	? NEURO
<input checked="" type="checkbox"/> Mitochondrial Encephalomyopathies	👍	👎	? NEURO
Paraneoplastic Neurologic Syndrome	👍	👎	? NEURO
Neuronal Ceroid Lipofuscinoses	👍	👎	○ NEURO
Why did this diagnosis come up ? ⓘ			
We matched the terms: myoclonic seizure myoclonic seizures dementia demential			
Degree of match between query entered and Isabel database (Not clinical probability): 23%			
Cerebral Amyloidosis	👍	👎	? NEURO
<input checked="" type="checkbox"/> Transient Ischemic Attack	👍	👎	? NEURO
CVA / Stroke	👍	👎	? NEURO
<input type="checkbox"/> Chorea Disorders	👍	👎	? NEURO
Huntington's Disease	👍	👎	? NEURO
<input checked="" type="checkbox"/> Multiple Sclerosis	👍	👎	? NEURO

feedback: [back to 10 >](#)