
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:

- **Age**: adult (30-39 yrs)
- **Gender**: female
- **Pregnancy**: (not specified)

Refine search:
- **Travel history**: North America

Enter clinical features, no negatives, no numbers:
- Myoclonic epilepsy
- Cognitive decline
- Memory loss
- Anosmia

+ add a clinical feature

**Diagnoses**

- **Nervous System Disorders**
  - Human Prion Diseases
  - Frontotemporal Dementia
  - HIV Neuro Complications
  - Mitochondrial Encephalomyopathies
  - Paraneoplastic Neurologic Syndrome
  - Neuronal Ceroid Lipofuscinoses

Why did this diagnosis come up?

We matched the terms: myoclonic seizure | myoclonic seizures | dement | demental

Degree of match between query entered and Isabel database (not clinical probability): 23%

- Cerebral Amyloidosis
- Transient Ischemic Attack
- CVA / Stroke
- Chorea Disorders
- Huntington's Disease
- Multiple Sclerosis

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