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**Case 17 (June 10 2010):** [N Engl J Med. 362\(23\):2213-2219](#)

**Demographic:** Female, 29 yrs, North America

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

weakness and flexion of left hand and ankle  
orofacial dystonia  
dysphagia  
difficulty speaking  
wide based gait  
falls  
anarthria

**Synonyms used by Isabel for the above query:**

dysphagia  
difficulty speaking

**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:**

Wilson's disease

**Final Diagnosis of the case according to NEJM:**

Rapid-onset dystonia-parkinsonism due to a mutation in the ATP1A3 gene

**Differential Diagnoses of the case as given by Isabel:**

Wilson's disease in Genetic

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**Was the final diagnosis given by Isabel:**

Yes, Rapid-onset dystonia-parkinsonism in Neuro

**Isabel 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: ⓘ

weakness and flexion of the left hand
orofacial dystonia
dysphagia
difficulty speaking
wide based gait
falls
anarthria

+ add a clinical feature

**get checklist** ➔

clear search

**diagnoses**

**Most Relevant**

sort by: most relevant | action: -select-

show: 10 20 30 all

**Rapid Onset Dystonia Parkinsonism**  NEURO

**Why did this diagnosis come up ?** ⓘ

We matched the terms: dysphagia | dysphagia to solids | orofac | orofacial | dystonia | fall | falls | anarthria

Degree of match between query entered and Isabel database (Not clinical probability): 59%

⊕ Muscular Dystrophies	? NEURO/MUSC
⊕ Dystonia Disorders	? NEURO
⊕ Hereditary Motor-Sensory Neuropathies	? NEURO/MUSC
⊕ Human Prion Diseases	? NEURO
Dermatomyositis / Polymyositis	? RHEUM
Intracranial Hemorrhage	? NEURO
Nephrogenic Systemic Fibrosis	? DERM
Corticobasal Degeneration	? NEURO
Systemic Sclerosis	? RHEUM
⊕ Polyneuropathy Disorders	? NEURO/MUSC
Sciatic Nerve Injuries	? NEURO
⊕ Transient Ischemic Attack	? NEURO
Sarcoidosis	? RESP