

Case 26 (August 24 2006): [N Engl J Med 2006;355:831-9](#)

Demographic: Female, 19 yrs, not pregnant, America North,
General speciality

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

pain, stiffness, and cramping of extremities worsened during the day

spasms in both legs

unable to walk

burning sensation in the feet

spasms in neck

tremors in arms and legs

anxiety and irritable

dystonic posturing

Synonyms used by Isabel (optional) for the above query:

none

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:

Secondary dystonias

Drug-induced tardive dystonias

Antipsychotic drugs: dopamine receptor–blocking older typical and newer atypical drugs

Anxiolytic drug: buspirone

Antidepressant agents: selective serotonin-reuptake inhibitors

Dopaminergic drugs: levodopa and dopamine agonists

Antiemetic drugs: metoclopramide

Antiseizure drugs: phenytoin, carbamazepine, gabapentin

Cerebral palsy

Wilson’s disease

Mitochondrial encephalopathies

Neuroacanthocytosis

Pantothenate kinase–associated neurodegeneration (Hallervorden–Spatz disease)

Fahr’s disease

Primary dystonias (15 types characterized by genetic features), two forms of which to consider in this patient:

DYT1: Early-onset torsion dystonia, due to mutation in torsin A gene at chromosome 9q34

DYT5: Dopa-responsive dystonia

DYT5a: autosomal dominant, due to mutation in *GCH1* gene at chromosome 14q22.1–22.2, encoding GTPCH1 protein

DYT5b: autosomal recessive, due to mutation in TH gene at chromosome 11p15.5, encoding tyrosine hydroxylase protein

Final Diagnoses of the case according to NEJM:

Dopamine-responsive dystonia (Segawa’s syndrome)

Differential Diagnoses of the case as given by Isabel:

Segawa’s syndrome – Isabel 1st page

Idiopathic torsion dystonia – Isabel 1st page

Cerebral palsy – Isabel 1st page

Fahr’s disease – Isabel 2nd page

Drug-induced Dystonia – Isabel 2nd page

Was the final diagnosis given by Isabel:

Yes, Isabel 1st page

Entire presentation cut and pasted into the Isabel query box:

Segawa's syndrome – Isabel 2nd page

Cerebral palsy – Isabel 2nd page

Fahr's disease – Isabel 2nd page

Wilson's disease – Isabel 2nd page

Drug-induced Dystonia – Isabel 3rd page

Isabel differential for dystonia :

NERVOUS SYSTEM DISORDERS ▾		
↳ Movement Disorders-Drugs	88%	R
↳ Neuroacanthocytosis Disorders	84%	R
↳ Dystonia Disorders	84%	R
↳ Metachromatic Leukodystrophy	81%	R
↳ Chorea Disorders	79%	R
↳ GM1 Gangliosidosis	79%	R
↳ GM2 Gangliosidosis	79%	R
↳ Corticobasal Degeneration	79%	R
↳ Cerebral Palsy	69%	R

ORTHOPEDIC DISORDERS ▾		
↳ Torticollis	82%	R

OPHTHALMOLOGY ▾		
↳ Leber Hereditary Optic Neuropathy	79%	R

HUMAN GENETICS ▾		
↳ Wilson's Disease	65%	R

INFECTIOUS DISEASES ▾		
↳ Measles Infection	63%	R

METABOLIC DISEASES ▾		
↳ Niemann-Pick Disease	41%	R
↳ Homocystinuria	40%	R
↳ Free Sialic Acid Storage Disease	40%	R

NERVOUS SYSTEM DISORDERS		
↳ Movement Disorders-Drugs	88%	R
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↳ Metachromatic Leukodystrophy	81%	R
↳ Chorea Disorders	79%	R
↳ GM1 Gangliosidosis	79%	R
↳ GM2 Gangliosidosis	79%	R
↳ Corticobasal Degeneration	79%	R
↳ Cerebral Palsy	69%	R
↳ Fahr's Disease	63%	R

Isabel differential for extracted features:

NERVOUS SYSTEM DISORDERS ▾		
Cerebral Palsy	52%	R
↳ Dystonia Disorders	38%	R
Paroxysmal Movement Disorders	25%	R
↳ Ataxic Disorders	24%	R
Spinal Cord Tumors	24%	R
Lumbar Disc Protrusion	24%	R
Lumbar Spondylosis	24%	R
Fahr's Disease	23%	R
↳ Myelitis	22%	R
↳ Movement Disorders-Drugs	22%	R
Epilepsy & Seizure Disorders	21%	R
Stiff-Person Syndrome	18%	R
GASTROINTESTINAL DISORDERS ▾		
Non Ulcer Dyspepsia	28%	R
METABOLIC DISEASES ▾		
↳ Acute Porphyria	28%	R
INFECTIOUS DISEASES ▾		
Lyme Disease	24%	R
Tetanus	18%	R
NUTRITIONAL DISORDERS ▾		
Thiamine Deficiency	21%	R
IMMUNOLOGIC SYSTEM ▾		
Eosinophilia-Myalgia Syndrome	43%	R
RHEUMATIC DISEASES ▾		
Fibromyalgia	32%	R
↳ Systemic Sclerosis	17%	R
NEUROMUSCULAR DISORDERS ▾		
↳ Polyneuropathy Disorders	31%	R
↳ Hereditary Motor-Sensory Neuropathies	28%	R
↳ Muscular Dystrophies	28%	R
Guillain-Barré Syndrome	27%	R
Dermatomyositis / Polymyositis	24%	R
ORTHOPEDIC DISORDERS ▾		
Torticollis	29%	R
CARDIAC DISORDERS ▾		
↳ Ischemic Heart Disease	28%	R
ENDOCRINE SYSTEM ▾		
Hyperthyroidism	24%	R
PSYCHOLOGIC DISORDERS ▾		
Rett Syndrome	♀ 23%	R
ALLERGIC DISORDERS ▾		
↳ Arthropod Allergy	16%	R