
**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%
- Neuroacanthocytosis Disorders: 84%
- Dystonia Disorders: 84%
- Metachromatic Leukodystrophy: 81%
- Chorea Disorders: 79%
- GM1 Gangliosidoses: 79%
  - GM2 Gangliosidosis: 79%
  - Corticobasal Degeneration: 79%
  - Cerebral Palsy: 69%

**ORTHOPEDIC DISORDERS**
- Torticollis: 82%

**OPHTHALMOLOGY**
- Leber Hereditary Optic Neuropathy: 79%

**HUMAN GENETICS**
- Wilson's Disease: 85%

**INFECTIOUS DISEASES**
- Measles Infection: 63%

**METABOLIC DISEASES**
- Niemann-Pick Disease: 41%
  - Homocystinuria: 40%
  - Free Sialic Acid Storage Disease: 40%
### Isabel differential for extracted features:

**NERVOUS SYSTEM DISORDERS**
- Cerebral Palsy: 52%
- Dystonia Disorders: 38%
- Paroxysmal Movement Disorders: 25%
- Ataxic Disorders: 24%
- Spinal Cord Tumors: 24%
- Lumbar Disc Protrusion: 24%
- Lumbar Spondylitis: 24%
- Fahr’s Disease: 23%
- Myelitis: 22%
- Movement Disorders-Drugs: 22%
- Epilepsy & Seizure Disorders: 21%
- Stiff-Person Syndrome: 18%

**IMMUNOLOGIC SYSTEM**
- Eosinophilia-Myalgia Syndrome: 43%

**RHEUMATIC DISEASES**
- Fibromyalgia: 32%
- Systemic Sclerosis: 17%

**NEUROMUSCULAR DISORDERS**
- Polyneuropathy Disorders: 31%
- Hereditary Motor-Sensory Neuropathies: 20%
- Muscular Dystrophies: 20%
- Guillain-Barré Syndrome: 27%
- Dermatomyositis / Polymyositis: 24%

**ORTHOEPEDIC DISORDERS**
- Torticollis: 23%

**CARDIAC DISORDERS**
- Ischemic Heart Disease: 26%

**ENDOCRINE SYSTEM**
- Hyperthyroidism: 24%

**PSYCHOLOGIC DISORDERS**
- Rett Syndrome: 23%

**ALLERGIC DISORDERS**
- Arthrosed Allergy: 18%