Case 35 (November 16 2006): N Engl J Med 2006;355:2132-42

Demographic: Male, Neonate, America North, General speciality

Clinical features extracted from the case and entered into the query box:
diffuse hypotonia
highpitched cry
mild jaundice
decreased tone in abdominal wall muscles
absent step and minimal Moro's reflexes
marked head lag, arms and legs held in a flaccid extensor posture
absent deep-tendon reflexes in the arms
trace reflexes at the knees
bilateral ankle clonus
polymicrogyria
elevated serum creatine kinase

Synonyms used by Isabel for the above query:
Highpitched cry

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

Final Diagnoses of the case according to NEJM:
Congenital muscular dystrophy

Differential Diagnoses of the case as given by Isabel:
Congenital muscular dystrophy (Muscular Dystrophies) – Isabel 1st page

Was the final diagnosis given by Isabel:
Yes, Isabel 1st page

Entire presentation cut and pasted into the Isabel query box:
None of the above

Isabel differential of extracted features from entire case presentation:

Table 1: Differential Diagnosis of the Floppy Infant Syndrome

Central nervous system disorders
Congenital, nonprogressive encephalopathies
Ischemic encephalopathies
Infectious encephalopathies
Metabolic encephalopathies
Endocrine encephalopathies
Developmental encephalopathies (e.g., Prader–Willi syndrome)
Degenerative, progressive encephalopathies

Spinal cord disorders (anterior horn cell and peripheral nervous system)
Infections (e.g., poliomyelitis)
Motor neuron diseases (spinal muscular atrophy type 1)
Neurogenic arthrogryposis
Glycogen storage diseases (e.g., Pompe's disease)
Lysosomal storage abnormalities
Sensorimotor polyneuropathies
Demyelinating disorders
Axonal disorders

Disorders of the neuromuscular junction
Presynaptic disorders
Infantile botulism
Congenital myasthenia
Postsynaptic disorders
Neonatal myasthenia gravis
Congenital myasthenia

Muscle disorders
Infantile myotonic dystrophy
Congenital myopathies

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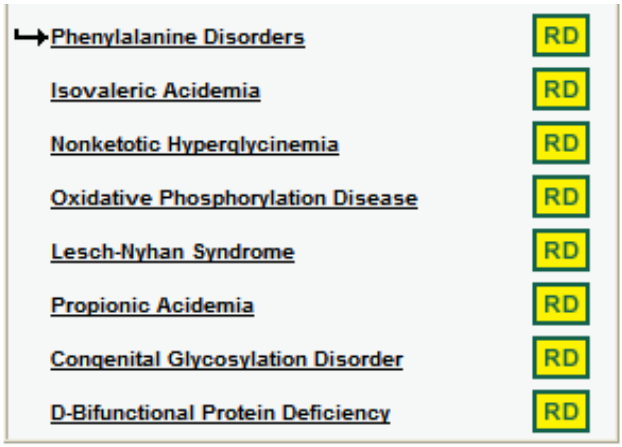
Isabel differential for floppy infant:


Phenylalanine Disorders RD
Isovaleric Acidemia RD
Nonketotic Hyperglycinemia RD
Oxidative Phosphorylation Disease RD
Lesch-Nyhan Syndrome RD
Propionic Acidemia RD
Congenital Glycosylation Disorder RD
D-Bifunctional Protein Deficiency RD