

Case 28 (September 11 2008): [N Engl J Med. 359\(11\):1156-1167](#)

Demographic: Male, 8 days old, America North, General speciality

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

congenital deafness
lethargy
hypothermia
weakness
respiratory distress
raised intracranial pressure
hypotonia
elevated lactate in CSF
abnormal posturing

Synonyms used by Isabel for the above query:

Lethargy
Respiratory distress
Raised intracranial pressure
Hypotonia
Abnormal posturing

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:

Mitochondrial encephalopathy
Bacterial Meningitis
Viral Meningoencephalitis – Herpes Simplex

Final Diagnosis of the case according to NEJM:

Leigh's syndrome

Differential Diagnoses of the case as given by Isabel:

Leigh's syndrome - under Nervous system disorders heading
Bacterial Meningitis – under Infectious diseases heading
Viral Meningoencephalitis – Herpes Simplex – under Infectious diseases heading

Was the final diagnosis given by Isabel:

Yes, under Nervous system disorders heading

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Isabel Differential:

+ INFECTIOUS DISEASES << click to expand	+ NERVOUS SYSTEM DISORDERS << click to expand
<input type="checkbox"/> Bacterial Meningitis ? RD <input type="checkbox"/> Viral Meningoencephalitis ? RD	<input type="checkbox"/> Epilepsy & Seizure Disorders ? RD <input type="checkbox"/> Mitochondrial Encephalomyopathies ○ RD
+ NEUROMUSCULAR DISORDERS << click to expand	<p>Why did this diagnosis come up ? ✖</p> <p>We matched the terms: seizures seizure respiratory distress hypotonia hypotonic hypothermia weak weakness elev elevated lactat lactate csf</p> <hr/> <p>Degree of match between query entered and isabel database (Not clinical probability): 65%</p> <ul style="list-style-type: none"> └ ATPASE Subunit 6 Mutation └ Kearns-Sayre Syndrome └ <u>Leigh's Disease</u> └ Thymidine Phosphorylase Deficiency
+ CONGENITAL MYOPATHIES << click to expand	
NEONATAL INFANT	
<input type="checkbox"/> Infant of a Diabetic Mother ? RD	
OBSTETRIC DISORDERS	
<input type="checkbox"/> Chorioamnionitis ? RD	
+ METABOLIC DISEASES << click to expand	
+ CARDIAC DISORDERS << click to expand	
+ NUTRITIONAL DISORDERS << click to expand	
	SHOCK STATES
	<input type="checkbox"/> Neonatal Sepsis ? RD
	ENDOCRINE SYSTEM
	<input type="checkbox"/> Hypothyroidism ? RD
	GASTROINTESTINAL DISORDERS
	<input type="checkbox"/> Hiatus / Diaphragmatic Hernia ? RD
	+ RESPIRATORY DISORDERS << click to expand
	+ HEMATOLOGY << click to expand
	+ LIVER DISORDERS << click to expand