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**Case 12 (April 21 2011):** [N Engl J Med. 364\(16\):1545-1556](#)

**Demographic:** Male, 9 months, North America

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

fever  
diarrhea  
microvesicular steatosis  
lethargy  
irritability  
intermittent vomiting  
elevated triglyceride level  
hyperammonemia

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

fever  
diarrhea  
lethargy  
irritability  
hyperammonemia  
elevated triglyceride level  
microvesicular steatosis

**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:**

Hemaphagocytic lymphohistiocytosis

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

Microvesicular hepatic steatosis due to CACT deficiency

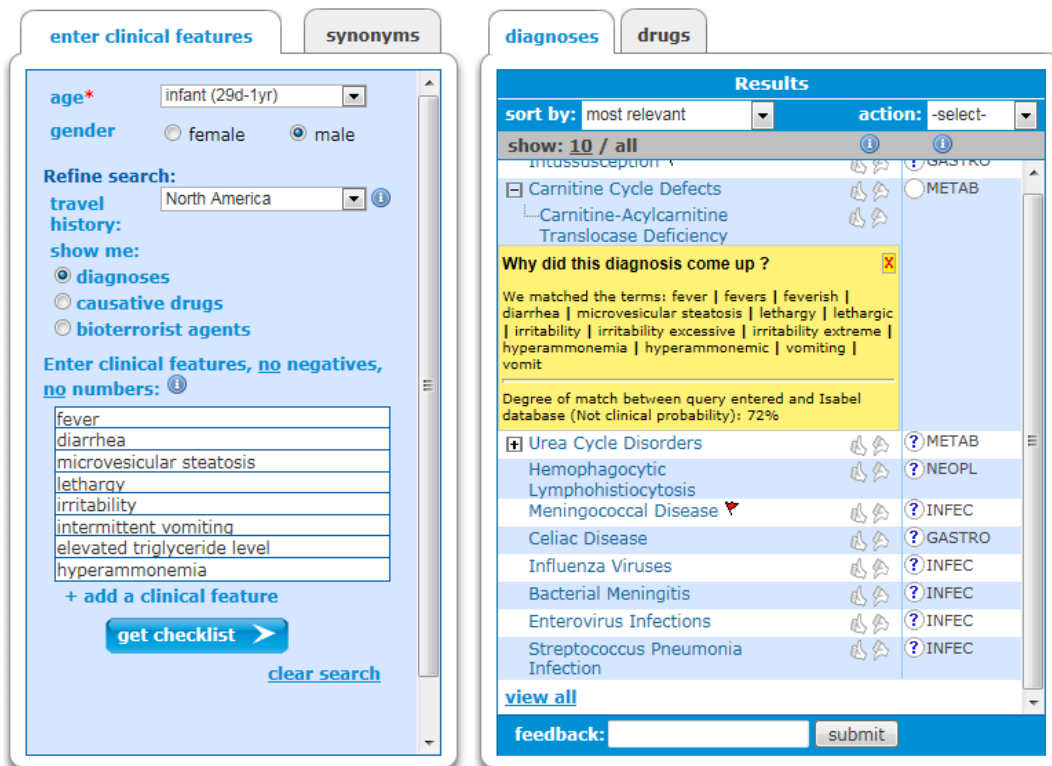
**Differential Diagnoses of the case as given by Isabel:**

Hemaphagocytic lymphohistiocytosis in Neoplastic

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

Yes, Carnitine-Acylcarnitine Translocase Deficiency in Metabolic

**Isabel Checklist Differential:**



The screenshot displays the Isabel healthcare interface, divided into two main panels: 'enter clinical features' and 'diagnoses'.

**enter clinical features panel:**

- age\*:** infant (29d-1yr)
- gender:** female (radio button), male (radio button, selected)
- Refine search:** travel history: North America
- show me:**
  - diagnoses
  - causative drugs
  - bioterrorist agents
- Enter clinical features, no negatives, no numbers:**
  - fever
  - diarrhea
  - microvesicular steatosis
  - lethargy
  - irritability
  - intermittent vomiting
  - elevated triglyceride level
  - hyperammonemia
- + add a clinical feature**
- get checklist** (button)
- clear search** (link)

**diagnoses panel:**

- Results** (header)
- sort by:** most relevant
- action:** -select-
- show: 10 / all**
- Why did this diagnosis come up ?** (highlighted section):
  - We matched the terms: fever | fevers | feverish | diarrhea | microvesicular steatosis | lethargy | lethargic | irritability | irritability excessive | irritability extreme | hyperammonemia | hyperammonemic | vomiting | vomit
  - Degree of match between query entered and Isabel database (Not clinical probability): 72%
- Urea Cycle Disorders** (category):
  - Hemophagocytic Lymphohistiocytosis (category: NEOPL)
  - Meningococcal Disease (category: INFECTION)
  - Celiac Disease (category: GASTRO)
  - Influenza Viruses (category: INFECTION)
  - Bacterial Meningitis (category: INFECTION)
  - Enterovirus Infections (category: INFECTION)
  - Streptococcus Pneumonia Infection (category: INFECTION)
- view all** (link)
- feedback:** (input field)
- submit** (button)



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