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**Case 3 (January 28 2010):** [N Engl J Med. 362\(4\):346-356](#)

**Demographic:** Male, 5 months, America North, General speciality

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

developmental delay  
excessive irritability  
hypotonia  
feeding problems

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

developmental delay  
excessive irritability  
hypotonia  
feeding problems

**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 PRO contain
- Did Isabel PRO suggest the Final diagnosis

**Differential Diagnoses considered by the MGH panel:**

Cerebral palsy

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

Infantile Krabbe disease (globoid-cell leukodystrophy)

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

Cerebral palsy – under Nervous system disorders

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

Yes, Krabbe disease under Metabolic disease

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### Isabel PRO Final Diagnosis:

**enter clinical features**      **synonyms**

**age** infant (28 days - 1 year) ▾  
**gender** male ▾  
**region** America North ▾ ⓘ  
**speciality** General ▾

Enter features, no negatives or numbers: ⓘ

developmental delay  
excessive irritability  
hypotonia  
feeding problems

+ add a feature  
**get checklist >>**

search again

Isabel PRO is not meant to replace your clinical judgment.

**relevance**      **commoner diagnoses**

**Diagnoses**

β-Oxidation Cycle Defects	? RD
3-Hydroxyisobutyryl-CoA Deacylase Deficiency	? RD
3-Methylcrotonyl-CoA Carboxylase Def	? RD
Galactose Metabolism Defects	? RD
Hepatocellular Failure	? RD
Hypoglycemia Disorders	? RD
Kabuki Syndrome	? RD
Krabbe Disease	○ RD

**Why did this diagnosis come up ?** ⓘ

We matched the terms: developmental delay | irritability | feeding problem | feeding problems

**Degree of match between query entered and Isabel database (Not clinical probability): 33%**

Nonketotic Hyperglycinemia	? RD
Renal Failure	? RD

show: 10 20 30 all

**10 Body Systems**

- Endocrine Disorders
- Nervous System Disorders
- Neuromuscular Disorders
- Nephrology Disorders
- Infectious Diseases
- Hematology Disorders
- Nutritional Disorders