
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
Isabel PRO Final Diagnosis:

Enter clinical features
- Age: infant (28 days - 1 year)
- Gender: Male
- Region: Amanda North
- Specialty: General

Enter features, no negatives or numbers:
- Developmental delay
- Excessive irritability
- Hypotonia
- Feeding problems

Add feature: get checklist

Isabel PRO is not meant to replace your clinical judgment.

Relevance

Diagnoses:
- 3-Oxidation Cycle Defects
- 3-Hydroxylisobutryl-CoA Deacylase Deficiency
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- Galactose Metabolism Defects
- Hepatocellular Failure
- Hypoglycemia Disorders
- Kabuki Syndrome
- Krabbe Disease
- Nonketotic Hyperglycinemia
- Renal Failure

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): 22%

10 Body Systems
- Endocrine Disorders
- Nervous System Disorders
- Neuromuscular Disorders
- Nephrology Disorders
- Infectious Diseases
- Hematology Disorders
- Nutritional Disorders