

Case 21 (July 14 2005): [N Engl J Med 2005;353:189-198](#)

Demographic: Male, 4 Week Old, America North, General speciality

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

jaundice
hyperbilirubinemia
thrombocytopenia
distended abdomen
icteric sclerae
palpable spleen
systolic ejection murmur
abnormal liver function

Synonyms used by Isabel for the above query:

distended abdomen
jaundice
thrombocytopenia
systolic ejection murmur
hyperbilirubinemia
palpable spleen

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:

Neonatal hemochromatosis

Final Diagnoses of the case according to NEJM:

Neonatal hemochromatosis

Neonatal giant cell hepatitis

Differential Diagnoses of the case as given by Isabel:

Neonatal hemochromatosis – Yes, 1st page

Was the final diagnosis given by Isabel:

Neonatal hemochromatosis – Yes, 1st page

Viral hepatitis – Yes, 1st page

Entire presentation cut and pasted into the Isabel query box:

Neonatal hemochromatosis - No

Neonatal giant cell hepatitis – Yes, click on Liver disorders heading

Isabel differential for extracted features:

LIVER DISORDERS << Click here ↳ <u>Cholestasis</u> RD <u>Physiologic Jaundice</u> RD ↳ <u>Viral Hepatitis</u> RD <u>Biliary Atresia</u> RD <u>Portal Vein Thrombosis / Occlusion</u> RD	SHOCK STATES << Click here <u>Neonatal Sepsis</u> RD
NEOPLASTIC DISEASES << Click here	INFECTIOUS DISEASES << Click here ↳ <u>CMV Infections</u> RD
UROLOGIC DISORDERS << Click here	HEMATOLOGY << Click here ↳ <u>Hemochromatosis</u> RD ↳ <u>Hemolytic Disease of the Newborn</u> RD <u>Autoimmune Hemolytic Anemias</u> RD
ENDOCRINE SYSTEM << Click here	↑ Neonatal Hemochromatosis
METABOLIC DISEASES << Click here	