WHAT'S BEHIND YOUR PATIENT'S HEMATOLOGICAL ABNORMALITIES?

RULE OUT MALIGNANCY. PERFORM GAUCHER ENZYME TEST.*

Patient Case Study 1

18-Year-Old Female Patient

HISTORY

- Admitted to hospital with a complaint of left-sided abdominal pain x 3 days
- No nosebleeds, blood in stool, trauma, weight loss, or bone pain
- Three weeks prior to admission: fatigue, fever, pharyngitis
- Parents both from Poland and of Ashkenazi Jewish origin

EXAM

- Pale
- Diffuse abdominal tenderness
- No bone/joint tenderness
- Hepatic margin 6 cm below costal margin

INVESTIGATIONS†

- Blood chemistries, urinalysis WNL
- Hb 104 g/L (120-160 g/L)
  PLT 27,000 x 10^9/L (150-450 x 10^9/L)
  Monospot negative
  WBC 3600/µl (4000-11,000/µl)
  PT 15.9 s (11-13 s)
  EBV titers consistent with past infection
  ANC 2016/µl (2000-8250/µl)
  aPTT 35.0 s

†Normal range in parentheses.

DIAGNOSTIC TEST

- Due to the clinical findings and family of origin, an enzyme assay (glucocerebrosidase) was ordered

DIAGNOSIS CONFIRMED: GAUCHER DISEASE TYPE 1

*Acid-ß-glucosidase enzyme activity assays.

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What's behind your patient's hematological abnormalities?

Patient Case Study 2

35-Year-Old Male Patient[1-3]

HISTORY

- Presented to PCP with petechial rash on lower extremities
- Sudden rectal hemorrhage episode 3 months prior; occasional nasal bleeding during childhood
- Labs revealed thrombocytopenia
- Referred to 2 specialists who ruled out cirrhosis, *H. pylori*, and autoimmune disease
- Referred to hematologist because of persistent rash and thrombocytopenia

EXAM

- Bruises on trunk and extremities; fading petechiae
- Spleen tip palpable on inspiration; liver not palpable

IMAGING: ABDOMINAL U/S[†]

- Splenomegaly 18 cm (<13 cm) with multiple echogenic foci up to 3.5 cm
- Hepatomegaly 18.1 cm (13.5-16.5 cm) with fatty infiltration
- Multiple small calculi in gallbladder

INVESTIGATIONS[†]

- HIV, EBV, HCV negative
- HGB, WBC, differential: Normal
- Platelets: 64x10^9/L (150-450 x10^9/L)
- Ferritin: 550 µg/L (15-475 µg/L)
- HDL cholesterol: 31 mg/dL (>40mg/dL)
- Cryoglobulin screen negative

[†]Normal range in parentheses.

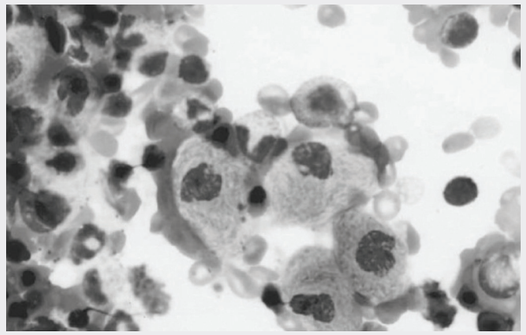
DIAGNOSTIC TEST

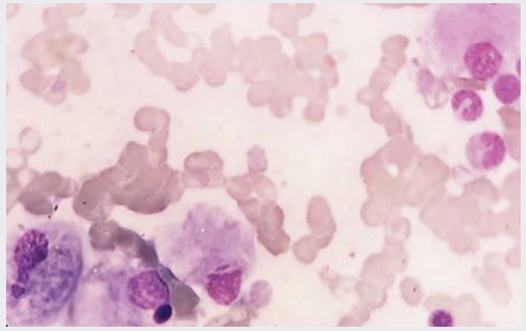
- Bone marrow biopsy ruled out malignancy
- Due to the clinical findings, an enzyme assay (glucocerebrosidase) was ordered

DIAGNOSIS CONFIRMED: GAUCHER DISEASE TYPE 1

Consider Gaucher disease in the differential.

Would you suspect Gaucher disease?





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Patient Case Study 1

18-Year-Old Female Patient

- Bone marrow biopsy:
  - Diffuse infiltration with Gaucher cells
  - No evidence of malignancy
- Enzyme assay confirmed Gaucher disease

Patient Case Study 2

35-Year-Old Male Patient

- Bone marrow biopsy:
  - Marked infiltration with Gaucher cells
- Enzyme assay confirmed Gaucher disease

- Bone marrow biopsy is not required for a Gaucher disease diagnosis. Not identifying Gaucher cells in bone marrow samples does not rule out Gaucher disease.[2]

IT'S GAUCHER DISEASE

Rule Out Malignancy

Gaucher disease type 1 commonly mimics the signs and symptoms of many hematological malignancies[2,4]

Test for It

Gaucher disease can be definitively diagnosed or ruled out with a simple blood-based enzymatic assay[2]

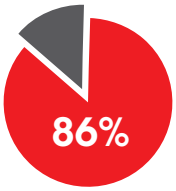
Treatable

Treatment options are available for Gaucher disease type 1, including oral therapies[3,5]

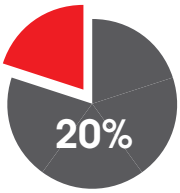
TEST TO KNOW

- Test for Gaucher disease in patients who present with splenomegaly and/or thrombocytopenia[2,4]
- Gaucher disease can be definitively diagnosed or ruled out with a simple blood-based enzymatic assay[4]

Hematologists/Oncologists Play a Pivotal Role in Diagnosis



of patients in the United States are seen by a hematologist/oncologist in pursuit of a diagnosis[4]



of hematologists/oncologists consider Gaucher disease in their differential diagnosis[4]

When the diagnosis is missed, a patient with Gaucher disease may experience a delay for up to 10 years[4]

In Non-Ashkenazi Patients[2]

Splenomegaly and/or thrombocytopenia?

YES

Hematological malignancies?

NO

Gaucher disease?

PERFORM GAUCHER ENZYME TEST*

Blood-based enzyme assay (acid beta-glucosidase) is the **gold standard** for definitive diagnosis of Gaucher disease[2]

In Patients of Ashkenazi Ancestry[2]

Splenomegaly and/or thrombocytopenia?

YES

NO

Low platelets?
Unexplained bleeding tendency?
Unexplained stable hyperferritinemia + normal transferrin saturation?
Increased inflammatory markers?

YES

Perform Gaucher Enzyme Test

Adapted from Mistry PK et al. *Am J Hematol.* 2011;86(1):110-115.

*Acid-ß-glucosidase enzyme activity assays.

In patients of Ashkenazi ancestry, the incidence of Gaucher disease (~1:850) is higher than hematologic malignancies (~1:2500). It is prudent to test for Gaucher disease as a first-line investigation in any patient of Ashkenazi ancestry presenting with splenomegaly and cytopenia.[2]

References: 1. Ito J, Saito T, Numakura C, et al. A case of adult type 1 Gaucher disease complicated by temporal intestinal hemorrhage. *Case Rep Gastroenterol.* 2013;7(2):340–346. doi: 10.1159/000354725 **2.** Mistry PK, Cappellini MD, Lukina E, et al. A reappraisal of Gaucher disease—diagnosis and disease management algorithms. *Am J Hematol.* 2011;86(1):110-115. doi:10.1002/ajh.21888 **3.** Pastores GM, Hughes DA. Gaucher disease. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews®*. University of Washington, Seattle. Published July 27, 2000. Updated June 21, 2018. Accessed July 14, 2022. https://www.ncbi.nlm.nih.gov/books/NBK1269/?report=printable **4.** Mistry PK, Sadan S, Yang R, Yee J, Yang M. Consequences of diagnostic delays in type 1 Gaucher disease: the need for greater awareness among hematologists-oncologists and an opportunity for early diagnosis and intervention. *Am J Hematol.* 2007;82(8):697-701. doi:10.1002/ajh.20908 **5.** Cox TM. Gaucher disease: clinical profile and therapeutic developments. *Biologics.* 2010;4:299-313. doi:10.2147/BTT.S7582

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