What’s behind your patient’s hematological abnormalities?

It may not be a malignancy.  It could be Gaucher disease.

### Patient Case Study 1

18-Year-Old Female

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

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

**INVESTIGATIONS**
- Blood chemistries, urinalysis WNL
- Hb 104 g/L  WBC 3600  ANC 2016
- PLT 27000  PT 15.9 s  PTT 35.0 s
- Monospot negative  EBV titers consistent w/ past infection

**DIAGNOSTIC TEST**
- Due to the clinical findings, an enzyme assay (glucocerebrosidase) was ordered

**DIAGNOSIS CONFIRMED: GAUCHER DISEASE TYPE 1**
HISTORY
› Patient referred because of skin rash and thrombocytopenia
› Rash had been present for 3-4 weeks, was erythematous and mostly macular, but not pruritic
› Patient told many years ago that spleen was palpable, and ~10 years ago that platelet count was low

EXAM
› Fading, mostly macular rash on trunk and extremities
› Spleen tip palpable on inspiration
› Liver not palpable
› No adenopathy

IMAGING: ABDOMINAL U/S*
› Splenomegaly 18cm (< 13cm) with multiple echogenic foci up to 3.5 cm
› Hepatomegaly 18.1cm (13.5 - 16.5cm) with fatty infiltration
› Multiple small calculi in gall bladder

INVESTIGATIONS*
› HGB, WBC, diff: Normal
› Platelets: 64x10⁹/L (150 – 450 x10⁹/L)
› Cryoglobulin screen +
› Serum protein electrophoresis: IgG-κ
› Paraprotein: 17 g/L or 1700 mg/dL
› Ferritin: 608 μg/L (15-475)
› ACE: 138 U/L (9-63)
› 24-hr urine protein: 0.25 g/dL (< .01 g/dL)*

DIAGNOSTIC TEST
› Due to the clinical findings, an enzyme assay (glucocerebrosidase) was ordered

DIAGNOSIS CONFIRMED: GAUCHER DISEASE TYPE 1

It May Not Be a Malignancy. It Could Be Gaucher Disease.

Rule Out Malignancy
Gaucher disease commonly mimics the signs and symptoms of many hematological malignancies¹²

Manageable
Early diagnosis and proper management are critical to preventing or reversing severe complications of Gaucher disease¹³
Consider Gaucher disease in the differential.

Would you suspect Gaucher disease?

Test to Know.

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

Patient Case Study 1

18-Year-Old Female

- Bone marrow biopsy:
  - Diffuse infiltration with Gaucher cells
  - No evidence of malignancy
- Enzyme assay confirmed Gaucher disease type 1

Patient Case Study 2

61-Year-Old Male

- Bone marrow biopsy:
  - Extensive infiltration with Gaucher cells
- Enzyme assay confirmed Gaucher disease type 1

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Hematologists Play a Pivotal Role in Diagnosis

Up to 86% of patients in the US are seen by a hematologist/oncologist in pursuit of a diagnosis

Only 20% of hematologists/oncologists consider Gaucher disease in their differential diagnosis

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

In Patients of Ashkenazi Ancestry

If splenomegaly and/or thrombocytopenia is present, perform Gaucher enzyme test.

NO

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

YES

Perform Gaucher Enzyme Test

In Non-Ashkenazi Patients

If splenomegaly and/or thrombocytopenia is present, perform Gaucher enzyme test.

YES Hematological malignancies? NO Gaucher Disease?

Perform Gaucher Enzyme Test

If you would like more information about identifying and managing patients with Gaucher disease, please contact Sanofi Genzyme Medical Information at 800-745-4447, option 2.