

# Take the Hem/Onc Diagnostic Challenge

19-year-old Portuguese-Irish male presented with low-normal hemoglobin, progressive **fatigue** and **thrombocytopenia**. Physical exam revealed a greatly enlarged spleen, confirmed by a CT Scan. Eight months after initial presentation he underwent a PET scan to rule out lymphoma. The results revealed a markedly **enlarged spleen** with uniform low-intensity uptake, but showed no indication of lymphoma or malignant neoplasms. One month later the patient underwent a **splenectomy**.

## Lab tests:

Hgb: 14.4 g/dL (13.5-18)  
 WBC:  $4.4 \times 10^3/\mu\text{L}$  (4-11)  
 Plts:  $59 \times 10^3/\mu\text{L}$  (150-400)  
 Neuts: 54% (45-70)  
 Lymphs: 35% (20-40)  
 Mono: 7% (3-10)  
 Eos: 3% (1-5)  
 Baso: 1% (0-0.5)



Normal Spleen



Splenomegaly

## The clinical challenge...

- What additional labs would you order?
  - Routine Chemistry Panel, SPEP, LDH
  - IEP, serum and urine, Beta 2 Microglobulin and Monospot
  - Peripheral blood smear, retic, haptoglobin, bone marrow aspiration/biopsy, Flow Cytometry
  - All of the above

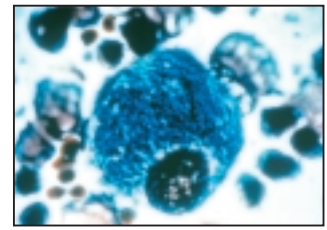
- Indicate your diagnosis from this list:

	<b>Likely</b>	<b>Unlikely</b>
A. Leukemia	—	—
B. Myelofibrosis	—	—
C. Gaucher disease	—	—
D. Thalassemia	—	—
E. Lymphoma	—	—
F. Infectious Mononucleosis	—	—
G. Chronic Hepatitis with Portal Hypertension	—	—

- What additional tests/information might confirm your diagnosis?
  - Angiotensin-Converting Enzyme (ACE), Chitotriosidase (Chito), Tartrate-Resistant Acid Phosphatase (TRAP)
  - Family History
  - Hepatitis Viral Studies

Their symptomatology suggested leukemia or lymphoma...

# ANSWER: Type I Gaucher disease



Gaucher cells

## DIAGNOSIS

The definitive diagnosis of Gaucher disease is made by enzyme assay of glucocerebrosidase in peripheral blood leukocytes.

Additional tests include:

- Gaucher serum markers: ACE, Chito, TRAP\*
- Abdominal MRI
- MRI of bilateral femurs
- CBC
- Ferritin level
- Lateral view of the entire spine
- Chemistry panel
- Complete radiological work-up
- DEXA scan

\*Gaucher disease biomarkers include Angiotensin-Converting Enzyme, Chitotriosidase, and Tartrate-Resistant Acid Phosphatase.

## KEY SIGNS AND SYMPTOMS

### 📌 Bone Impact:

96% of all Gaucher patients show radiologic signs of bone disease and are at high risk for osteopenia, osteoporosis and osteonecrosis



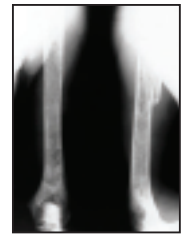
Bone marrow infiltration



Osteonecrosis



Osteopenia/pathologic fracture



### 📌 Visceral Impact:

- Spleen may enlarge up to 25 times normal size (splenectomy typically NOT recommended)
- Liver may enlarge up to 2.5 times normal size

### 📌 Hematologic Impact:

Thrombocytopenia, anemia, easy bruising and bleeding

## TREATMENT

**Type 1 Gaucher disease** is the most common lysosomal storage disorder with onset of symptoms at any age, from infancy to adulthood. **A referral to a physician knowledgeable about Gaucher disease or to a lysosomal storage disease center** is recommended.

## LEARN MORE

<http://www.gaucherregistry.org> for managing and monitoring guidelines

<http://www.geneclinics.org> for testing information

<http://www.gaucherdisease.org> for the National Gaucher Foundation

<http://www.genzyme.com> or call Genzyme Medical Information at 1-800-745-4447, option 2

**Early diagnosis and intervention are key – hematologists/oncologists can play a role.**

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