

# Diagnostic algorithm for investigation of possible Gaucher's disease in adults.

## **BE AWARE:**

*84% of Gaucher's patients have **high ferritin** levels,  
60% have **thrombocytopenia**,  
40-50% have **enlarged liver/spleen**, at diagnosis*

### ▪ **SPLENOMEGALY/HEPATOMEGALY and:**

- ✓ No evidence of portal hypertension
- ✓ Anaemia and/or Thrombocytopenia
- ✓ No definite evidence of blood malignancy
- ✓ MGUS/ polyclonal Ig increase in patients < 30 years
- ✓ Unexplained bone pain

### ▪ **THROMBOCYTOPENIA and:**

- ✓ Unexplained high ferritin with iron binding < 50%
- ✓ Normochromic normocytic anaemia; Hb < 110 in women, < 120 g/l in men
- ✓ Inconclusive investigations
- ✓ MGUS/ polyclonal Ig increase in patients < 30 years
- ✓ Unexplained bone pain

### ▪ **UNEXPLAINED HIGH FERRITIN LEVELS and IRON BINDING < 50%**

### **CONSIDER TESTING IN:**

- ✓ Unexplained aPTT prolongation
- ✓ High ACE levels without evidence of sarcoidosis

### **LEUCOCYTE ACID B-GLUCOSIDASE ASSAY**

- 1 BLUE and 1 GREEN TOP TUBES F.A.O. DUTY BIOCHEMIST
- SERVICE AVAILABLE MON-THU
- CENTRALISED LAB TESTING WITH NO COSTS TO THE TRUST

**IF POSITIVE, REFER PATIENT TO THE NEAREST LYSOSOMAL STORAGE DISEASE CENTRE, FOR HEFT:**

**Drs T. HIWOT/ C. DAWSON, INHERITED METABOLIC DISORDER UNIT  
QUEEN ELIZABETH HOSPITAL, FAX: 0121 371 6990**