

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

- **2 PURPLE 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:

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