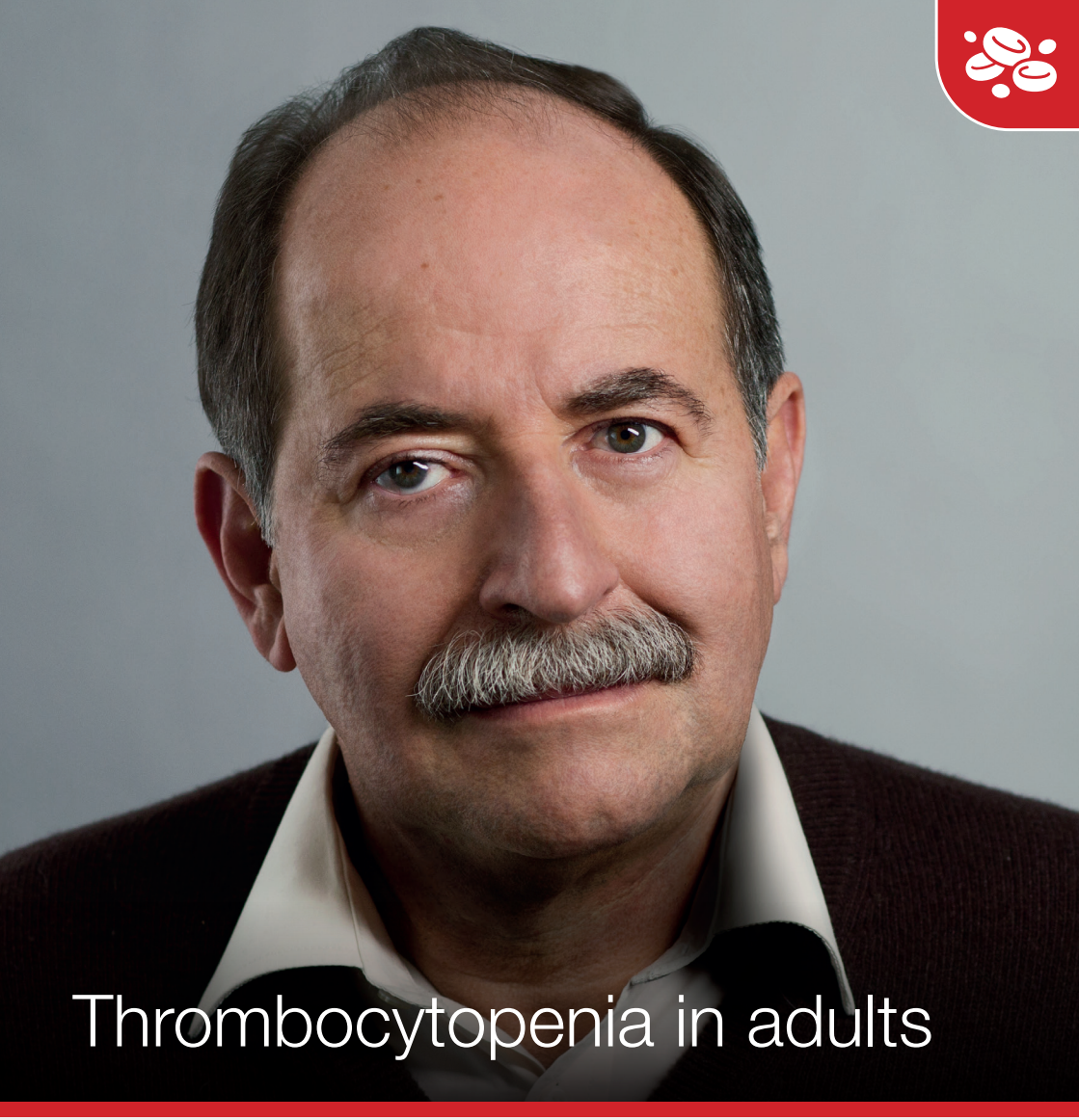


Thrombocytopenia in adults

Have you ruled out Gaucher disease?

Takeda

Gustavo has type 1 Gaucher disease

Have you considered Gaucher disease as a possible cause of thrombocytopenia?

Thrombocytopenia
Platelet count
$<150 \times 10^9$/L for adults[1,2]

A finding of thrombocytopenia during routine evaluation is a relatively common reason for referral to a haematologist[1]

May result from a wide range of conditions and may be determined by multiple mechanisms[1]

A common early feature in adults and children with type 1 Gaucher disease[3] (affecting **82% of patients** in one study[4])

Gaucher disease

Gaucher disease is an uncommon, chronic and progressive disorder, which can lead to disabling, irreversible complications and reduced life expectancy.[5,6]

Deficiency of the lysosomal enzyme glucocerebrosidase results in accumulation of its main substrate, glucosylceramide, and leads to cellular dysfunction.[7]

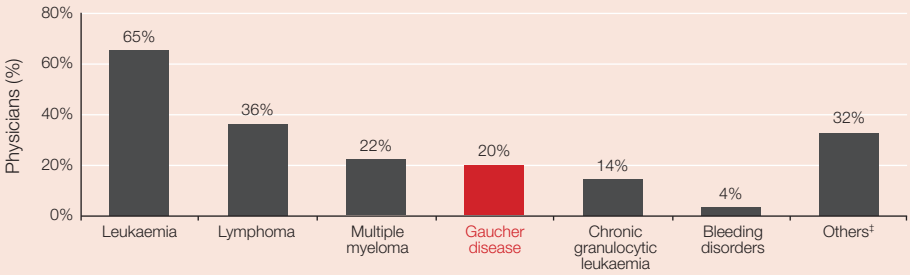
Classical symptoms:[8]

- **Cytopenia**
- **Hepatosplenomegaly**
- **Bone pain**

It is now known that **hyperferritinaemia*** is also common in Gaucher disease (87% of patients in one study)[4]

Many don't see Gaucher disease even when all the signs are there[8]

Differential diagnoses considered for a hypothetical case with classical symptoms† of Gaucher disease



Data from Mistry P, et al. 2007.[8]

*Elevated ferritin levels. †Cytopenia, hepatosplenomegaly and bone pain. ‡Anaemia, bone marrow failure/disease, cancer (unspecified), depository/storage disease (unspecified), fibrosis, haematological disease (unspecified), hepatitis/liver disease, Hodgkin disease, Legg-Calvé-Perthes disease, malaria, metastatic cancer, myelofibrosis, myeloma, myeloproliferative disorder, neoplasia, thalassaemia

3 STEPS to a differential diagnosis of thrombocytopenia in the general population

STEP 1: Exclude common and non-cancer causes[1]

- Family history/primary immune thrombocytopenia
- Drug-induced (e.g. heparin) thrombocytopenia
- Pregnancy
- Infections
- Connective tissue disorders

STEP 2: Exclude malignancy[1,9]

- Leukaemia
- Lymphoma

Patients with MGUS, myeloma or B-cell malignancy may also have concurrent Gaucher disease[10]

STEP 3: Consider checking for Gaucher disease*

Consider checking for Gaucher disease **if thrombocytopenia is present along with 1 or more red flags**, particularly if one is splenomegaly:[3,10]

Type 1 Gaucher disease

- **Splenomegaly**
- Hyperferritinaemia
- History of bone pain
- Family history of Gaucher disease or Ashkenazi Jewish ethnicity or Parkinson's disease

Type 3 Gaucher disease

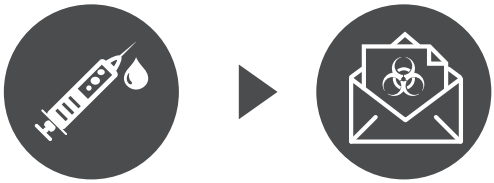
As for type 1 but also:

- Slow horizontal saccades with unimpaired vision, oculomotor apraxia
- Spinal deformities (gibbus)
- Myoclonic epilepsy

*Gaucher disease should be an initial consideration in any person of Ashkenazi ancestry presenting with splenomegaly or (if absent) thrombocytopenia (even if mild), bleeding tendency, unexplained stable hyperferritinaemia with normal transferrin saturation, or increased inflammatory markers[11]
MGUS, monoclonal gammopathy of undetermined significance

Gaucher disease can be ruled out using an enzyme assay

The presence of Gaucher disease can be tested using a widely available assay in peripheral white blood cells, to check for glucocerebrosidase enzyme activity.[10] **There is no need for bone marrow biopsy.[11]**

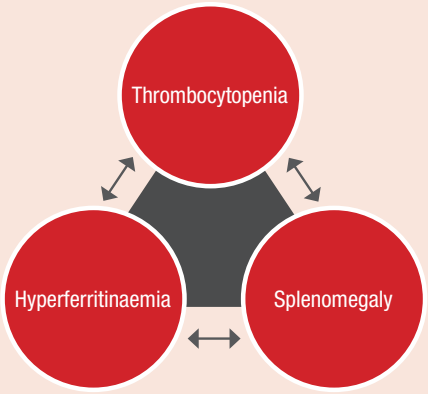


Collect a blood sample from your patient

Send for analysis

Takeda provides glucocerebrosidase enzyme testing, as a service to medicine. Please speak to a member of the Takeda Diagnostic Support Team for more information.

Gaucher disease – a triad of associated symptoms[3,4]



When you see these symptoms, rule out Gaucher disease

References: 1. Stasi R. Hematology Am Soc Hematol Educ Program. 2012;2012:191–197. 2. Biino G, *et al.* PLoS One. 2013;8(1):e54289. 3. Mehta A, *et al.* Intern Med J. 2019;49:578–591. 4. Thomas AS, *et al.* Blood Cells Mol Dis. 2013;50(3):212–217. 5. Weinreb NJ, *et al.* Am J Hematol. 2008;83:896–900. 6. Biegstraaten M, *et al.* Blood Cells Mol Dis. 2018;68:203–208. 7. Grabowski GA. Lancet. 2008;372:1263–1271. 8. Mistry P, *et al.* Am J Hematol. 2007;82(8):697–701. 9. Shahrabi S, *et al.* Histol Histopathol. 2018;33(9):895–908. 10. Weinreb NJ, *et al.* Blood. 2018;131(22):2500–2501. 11. Mistry P, *et al.* Am J Hematol. 2011;86(1):110–115.

C-ANPROM/UKI/GAUD/0023 | Date of preparation: December 2020.