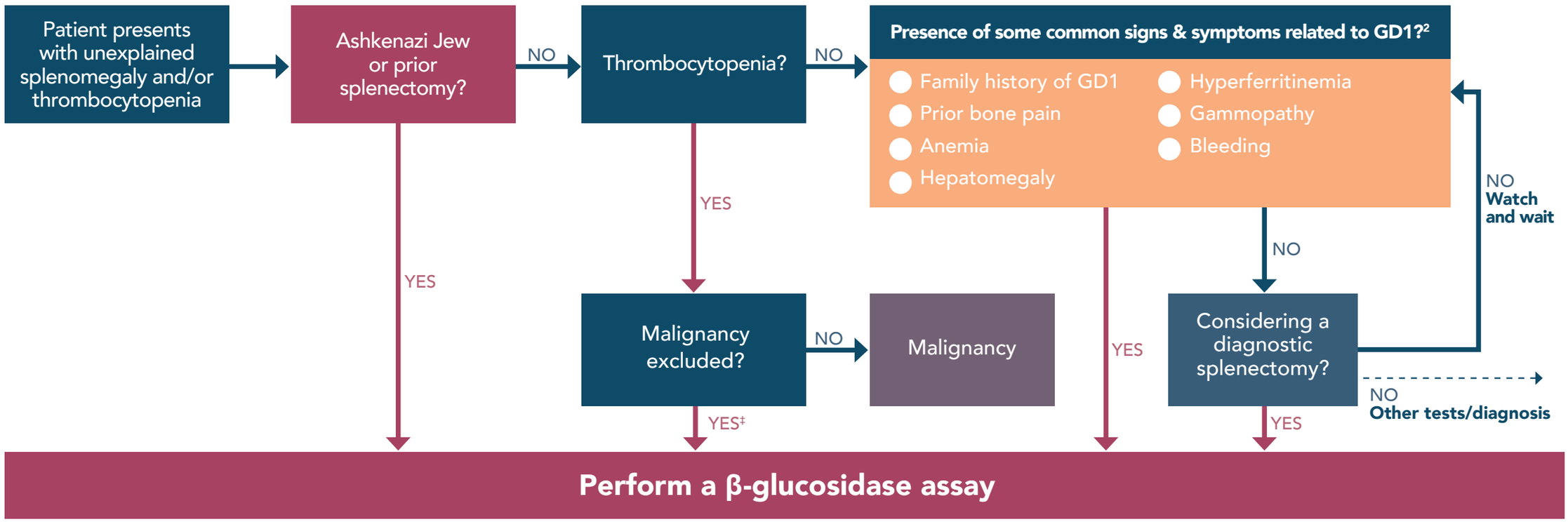
# A PROPOSED TYPE 1 GAUCHER DIAGNOSIS ALGORITHM FOR ADULT PATIENTS WITH UNEXPLAINED SPLENOMEGALY AND/OR THROMBOCYTOPENIA[1,2]

This proposed diagnosis algorithm provides information to assist in the differential diagnosis of type 1 Gaucher disease (GD1) in patients with unexplained splenomegaly and/or thrombocytopenia.*† The algorithm has been synthesized from two publications on GD1 diagnosis: *'Consensus Conference: A reappraisal of Gaucher disease - diagnosis and disease management algorithms'*[1] and *'Presenting signs and patient co-variables in Gaucher disease: outcome of the Gaucher Earlier Diagnosis Consensus (GED-C) Delphi initiative.'*[2]
**This algorithm is not intended to be a diagnostic tool. It does not replace the need for a complete evaluation of the patient by a healthcare professional.**




## SYMPTOMATIC FEATURES OF GD1:

| Feature[1,2] | Definition[2,3] | Symptoms[4] |
|---|---|---|
| Splenomegaly | Higher than spleen volume of an unaffected person (> 0.2% body weight) | Early satiety, abdominal pain |
| Thrombocytopenia | Lower than platelet count of an unaffected person (< 150 × $10^9$/L) | Bruising, nosebleeds,[5] post-operative bleeding |
| Anemia | Lower than hemoglobin levels of an unaffected person (< 140 g/L) | Fatigue |
| Hepatomegaly | Higher than liver volume of an unaffected person (> 2.5% body weight) | Early satiety, abdominal pain |

## NOTES:

*Splenomegaly and/or thrombocytopenia for which healthcare providers have found no medical cause, or whose cause remains contested.

† Portal hypertension due to advanced liver disease from well-known etiologies must be ruled out before application of the algorithm.

‡ **Immune thrombocytopenia (ITP)**
Splenomegaly provides evidence against a diagnosis of ITP.[6] Further, spontaneous bruising is uncommon in ITP unless the platelet count is less than 30 × $10^9$/L.[7]



**REFERENCES: 1.** Mistry *et al.* Am j hematol. 2011; 86(1):110–15 **2.** Mehta *et al.* Intern med j. 2019; 49(5):578–91 **3.** Pastores *et al.* Semin hematol. 2004; 41(4 Suppl 5):4–14. **4.** Thomas *et al.* Br j haematol 2014; 165(4):427–40
**5.** Nagral. Clin exp hepatol. 2014; 4(1):37–50 **6.** George *et al.* Blood. 1996; 88(1):3–40 **7.** Thomas *et al.* Blood cells mol dis. 2013; 50(3):212–17