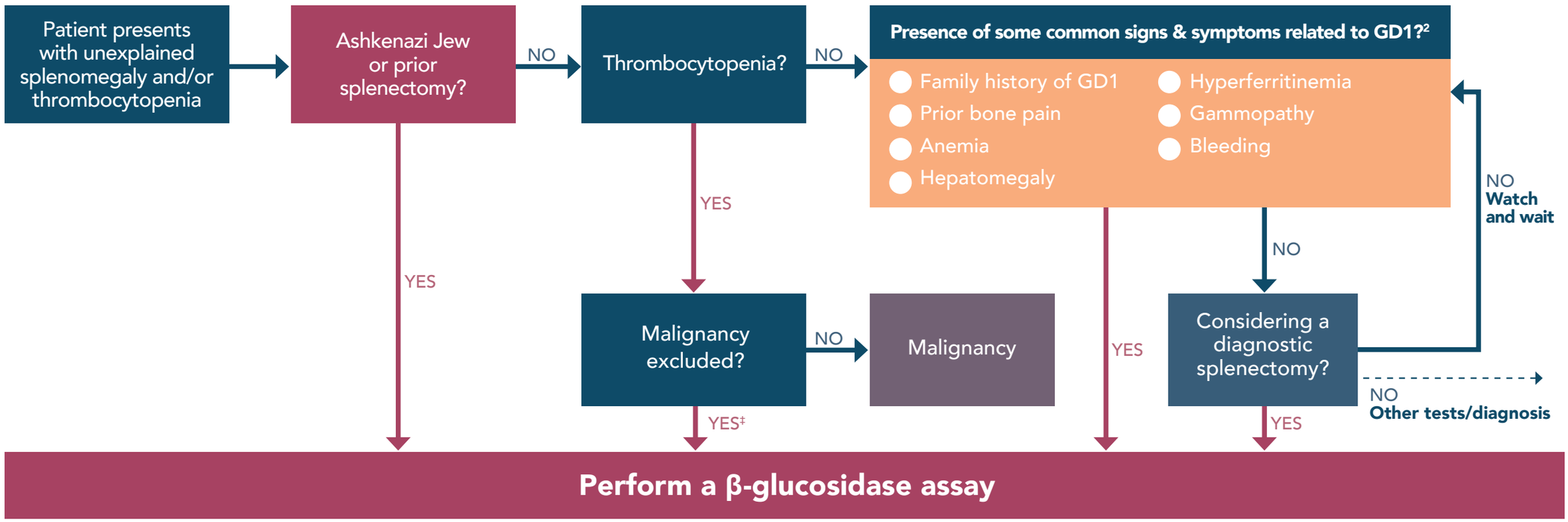
A PROPOSED TYPE 1 GAUCHER DIAGNOSIS ALGORITHM FOR ADULT PATIENTS WITH UNEXPLAINED SPLENOMEGALY AND/OR THROMBOCYTOPENIA[1,2]

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]

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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