KNOW RARE DISEASE. GAUCHER.

TYPE 1 GAUCHER DISEASE

DIAGNOSIS AND MONITORING

Clear, actionable steps for Healthcare providers—with a patient resource to share

Takeda

WHAT IS TYPE 1 GAUCHER DISEASE?

Type 1 Gaucher disease (GD1) is a rare genetic lysosomal storage disorder in which the enzyme glucocerebrosidase is deficient.[1–3] This leads to an accumulation of the fatty substance glucocerebroside in the affected cells and the formation of 'Gaucher cells.' Over time, Gaucher cells infiltrate various organs including the spleen, liver, and bone marrow, resulting in progressive organ damage.[2,4]

PREVALENCE

GD1 affects males and females equally and can affect any population, but incidence is much higher in people of Ashkenazi Jewish ancestry.[5,6] GD1 is an autosomal disorder.[6]

GD1 affects:

- 1–9 in 100,000 within the overall population[7]
- ~1 in 600 within the Ashkenazi Jewish population[5]
- ~1 in 17 within the Ashkenazi Jewish community who are carriers[8]
- An estimated 6,000 individuals in the United States[5,9]

SYMPTOMATIC FEATURES OF GD1

Feature[10,11]	Definition[11,12]	Symptoms[13]
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[6], 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
Skeletal manifestations	Any bone or bone-marrow abnormality attributable to accumulation of Gaucher cells in bone marrow[14]	Bone pain and fractures, osteonecrosis[6]

Early diagnosis is critical for managing and slowing disease progression of GD1, yet patients frequently experience considerable diagnostic delays due to its rarity, heterogeneity, and the overlap of GD1 symptoms with several other diseases.[15] The differential diagnosis for hematologic manifestations in GD1 includes other blood disorders, with leukemia, lymphoma, and multiple myeloma being common misdiagnoses due to symptoms like anemia, thrombocytopenia, and splenomegaly.[16]

Early diagnosis of GD1 is important due to the progressive nature.[2]

To learn more about GD1, and view other useful resources, visit **knowgaucherdisease.com/hcp**

MONITORING AND MANAGEMENT

Disease monitoring and management are critical for assessing and slowing the progression of GD1.[15] Current therapeutic goals reflect increased knowledge of potential treatment effects and include early detection of liver and other complications, improvement in mobility, and enhancement of quality-of-life measures such as fatigue and social participation.[15]

DISEASE MONITORING

The International Collaborative Gaucher Group (ICGG), a group of physicians with experience in the management of Gaucher disease established in 1991, has developed comprehensive guidelines for the clinical monitoring of adult patients with GD1. The ICGG pooled data from a large observational database registry to create these guidelines for comprehensive evaluation of patients.[17]

BLOOD TESTS

Blood tests include hemoglobin count and platelet count. Additionally, the serum levels of several biological markers indicate the severity of GD1, and disease progression[3,4,15]:

- Glucosylsphingosine (**lyso-Gb1/lyso-GL1**)[18]
- Chitotriosidase (**CHIT1**)[17,18]
- Chemokine ligand 18 (**CCL-18**)[17,18]
- Tartrate-resistant acid phosphatase (**TRAP**)[17]
- Angiotensin-converting enzyme (**ACE**)[17]

VISCERAL VOLUME

Visceral volumes can be assessed with:

- **Volumetric MRI** (since repeat assessment is routine in GD1)[17,18]
- **CT** or **ultrasound** where MRI is unavailable[17,18]

SKELETAL SCANS[1]

Bone marrow infiltration and bone disease can be assessed with:

- **MRI**, particularly bone infarction and necrosis[17]
- **DEXA**, the gold standard method for assessing BMD[17]

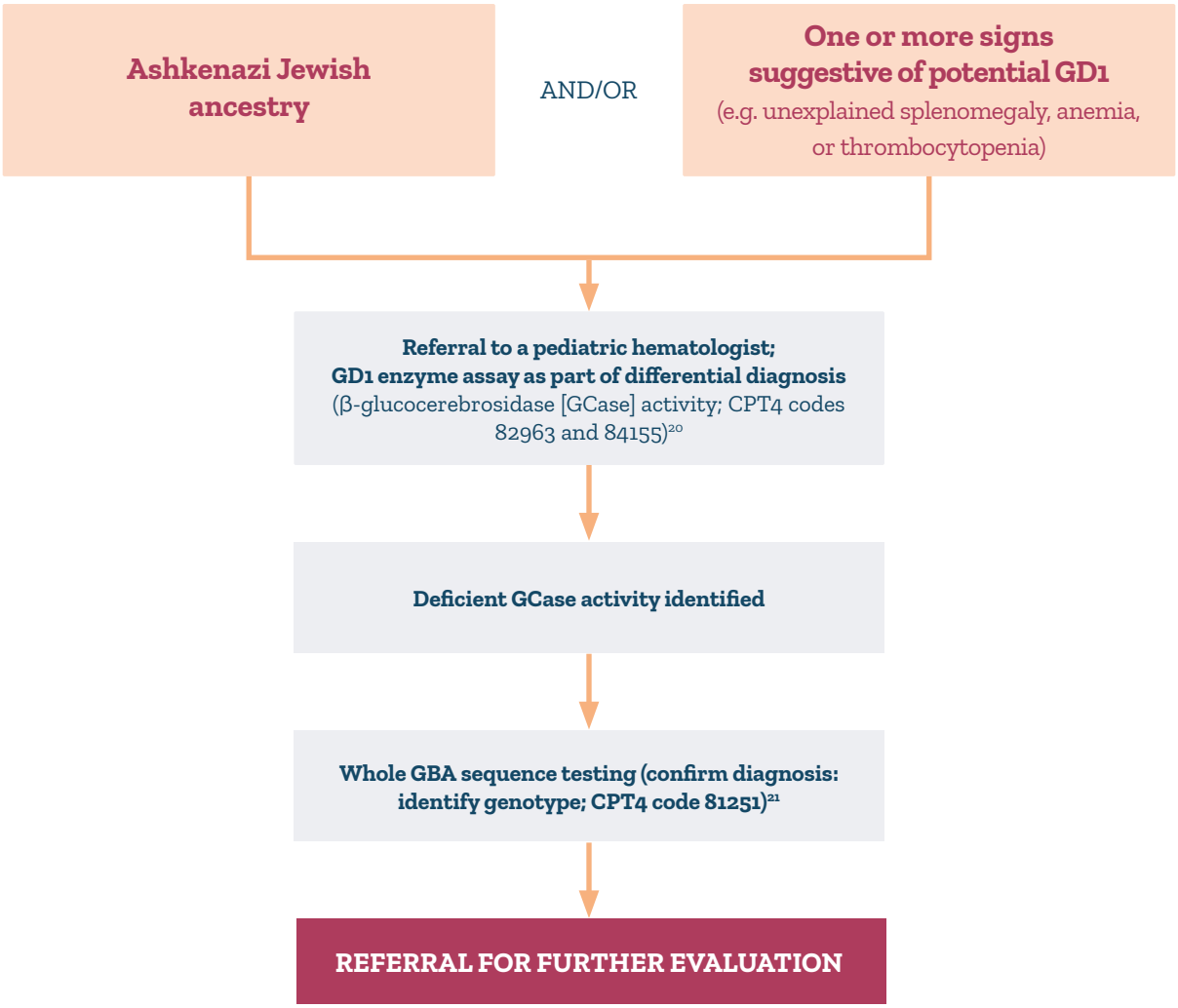
Discuss GD1 monitoring with your patients and explain how it is crucial for disease management.

BMD, bone mineral density; CT, computed tomography; DEXA, dual-energy X-ray absorptiometry; MRI, magnetic resonance imaging.

PEDIATRIC DIAGNOSTIC ALGORITHM

The following algorithm has been proposed for pediatric patients without a family history of GD1, based on a Delphi consensus.[15,19]

To assist in diagnosis, algorithms have been developed based on recent evidence and current understanding of biomarkers. These algorithms are not intended to be diagnostic tools. They do not replace the need for a complete evaluation of the patient by a healthcare professional.



Adapted from Weinreb et al. 2022

GD1 is diagnosed using a simple blood test measuring enzyme levels, which can be ordered through local labs.[6] A bone marrow biopsy is not diagnostic for Gaucher.[6]

ONGOING MONITORING FOR PEDIATRIC PATIENTS

	At baseline	Every 6–12 months	Every 12–24 months	Every 24–36 months	At time of dose change or significant complication
Physical examination Including growth measurements (height and weight)	✓	✓			✓
Hematology — Hemoglobin and platelets	✓	✓			✓
Hematology — PT and PTT in patients with bleeding symptoms	✓	✓			
Visceral Spleen and liver volumes	✓	✓			
Biomarkers Focus on lyso-Gb1	✓	✓			✓
Skeletal — MRI	✓		✓		
Skeletal — DEXA	✓			✓	
Pain and QoL QoL assessed by SF-36, PedsQL, or Kidscreen	✓	✓			

Adapted from Weinreb et al, 2022. DEXA, dual-energy X-ray absorptiometry; lyso-Gb1, glucosylsphingosine; MRI, magnetic resonance imaging; PedsQL, Pediatric Quality of Life Inventory; PT, prothrombin time; PTT, partial thromboplastin time; QoL, quality of life; SF-36, 36-item Short-Form Health Survey.

MANAGEMENT OF PEDIATRIC PATIENTS

TREATMENT

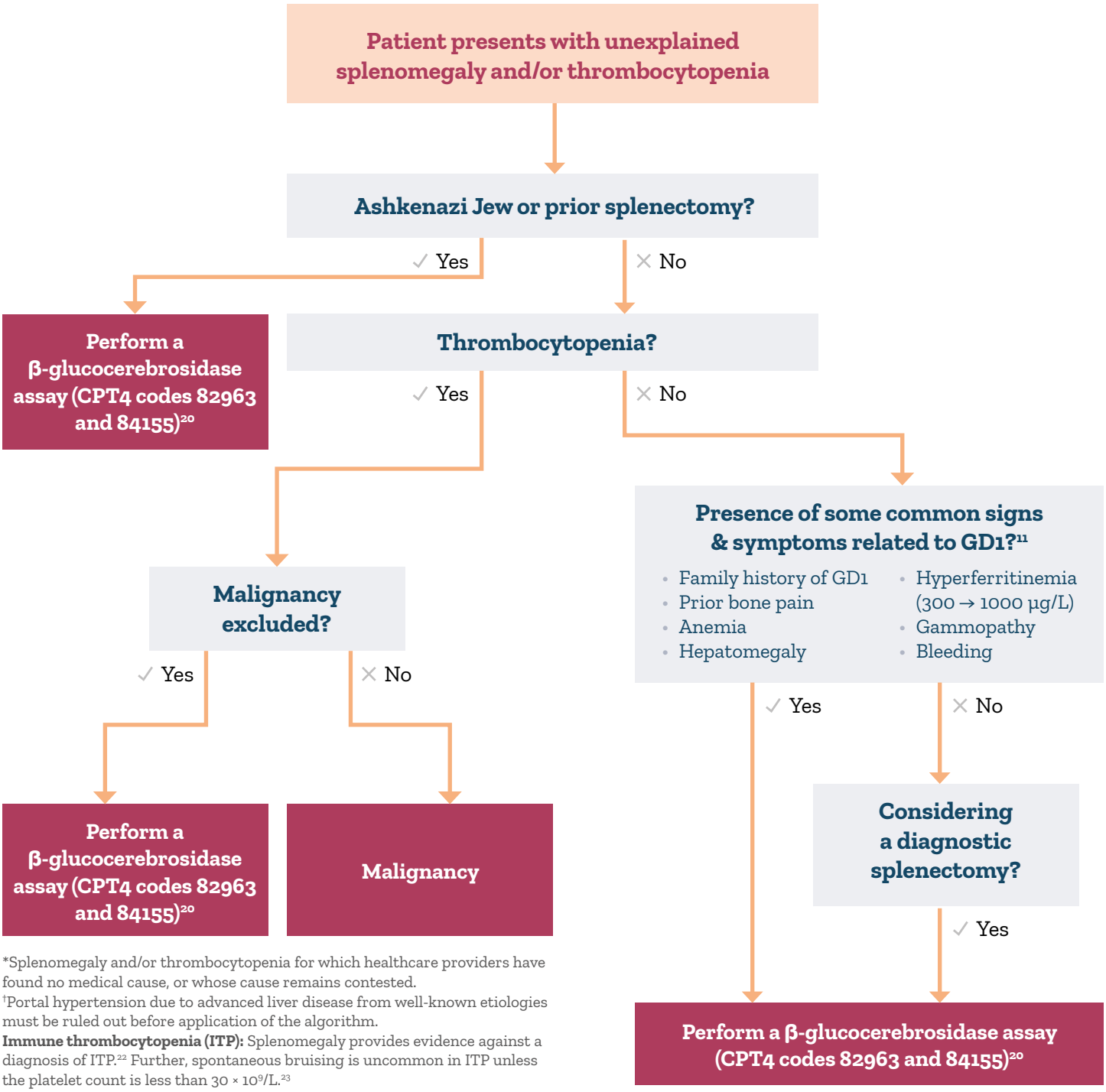
Regular intravenous infusions with enzyme-replacement therapy (ERT) are recommended for all children or adolescents with symptomatic GD1. ERT is currently the only FDA-approved treatment for GD1 in the pediatric population.[15]

THERAPEUTIC GOALS

Therapeutic goals involve: controlling thrombocytopenia, bleeding, and anemia; improving bone health and mobility; preventing or improving pulmonary and spleen complications; achieving normal growth and improving general well-being.[15]

ADULT DIAGNOSTIC ALGORITHM

This proposed diagnosis algorithm provides information to assist in the differential diagnosis of 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'[10] and 'Presenting signs and patient co-variables in Gaucher disease: outcome of the Gaucher Earlier Diagnosis Consensus (GED-C) Delphi initiative.'[11]



*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.[22] Further, spontaneous bruising is uncommon in ITP unless the platelet count is less than 30 × 10^9/L.[23]

ONGOING MONITORING FOR ADULT PATIENTS

NOT ON ENZYME REPLACEMENT[12,17]

Assessment	Every 12 months	Every 12–24 months
Comprehensive physical examination and SF-36* survey	✓	
Blood tests	✓	
Visceral volume		✓
Skeletal scans		✓

ENZYME REPLACEMENT: DID NOT ACHIEVE THERAPEUTIC GOALS[17]

Assessment	At 3 months	At 6 months	At 9 months	At 12 months
Comprehensive physical examination and SF-36* survey				✓
Blood tests	✓	✓	✓	✓
Visceral volume				✓
Skeletal scans				✓

ENZYME REPLACEMENT: ACHIEVED THERAPEUTIC GOALS[17]

Assessment	Every 12 months	Every 12–24 months
Comprehensive physical examination and SF-36* survey	✓	
Blood tests		✓
Visceral volume		✓
Skeletal scans		✓
Repeat schedule if patient has not achieved therapeutic goals		

*SF-36, 36-item Short-Form Health Survey. The ICGG has pooled data from patients in a large observational database registry.[17] The registry is intended to explore and define the natural history of GD1 and characterize patient response to therapy.[17] With the registry information, ICGG has developed a comprehensive evaluation for regular clinical monitoring that is dependent on the circumstances of the patient.[17]

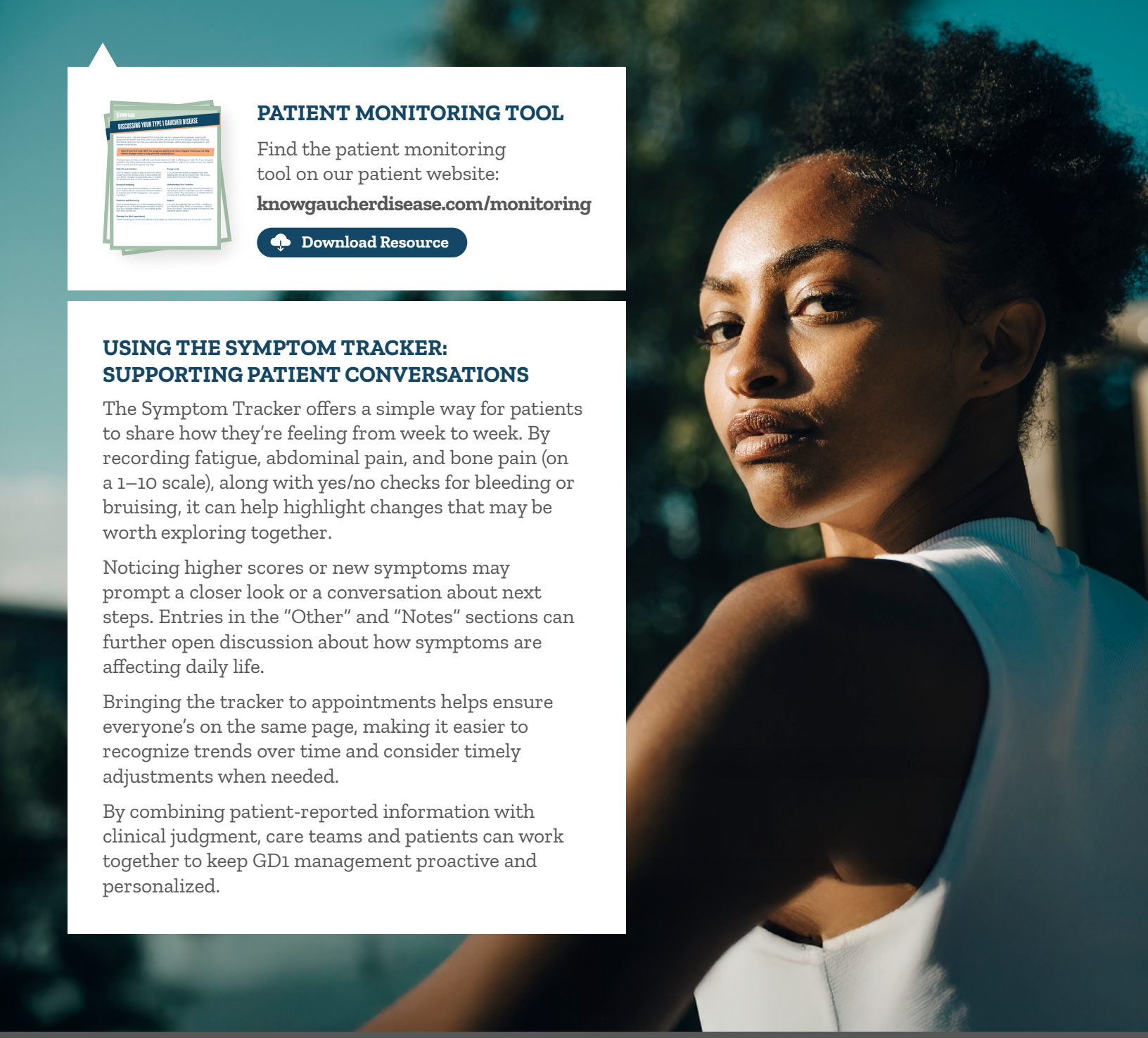
MANAGEMENT OF ADULT PATIENTS

TREATMENT

Treatment should be initiated immediately in patients with GD1 who have severe and/or progressive disease.[19] ERT should be recommended for patients who manifest severe and/or progressive signs and symptoms.[19] Substrate reduction therapy (SRT) may be used for eligible adults with GD1.[19]

THERAPEUTIC GOALS

Therapeutic goals involve: normalizing hemoglobin and platelet levels; maintaining bone health and mobility; reducing and maintaining liver and spleen sizes, and preventing complications; improving and preserving pulmonary function.[19]



PATIENT MONITORING TOOL

Find the patient monitoring tool on our patient website:

knowgaucherdisease.com/monitoring

Download Resource

USING THE SYMPTOM TRACKER: SUPPORTING PATIENT CONVERSATIONS

The Symptom Tracker offers a simple way for patients to share how they're feeling from week to week. By recording fatigue, abdominal pain, and bone pain (on a 1–10 scale), along with yes/no checks for bleeding or bruising, it can help highlight changes that may be worth exploring together.

Noticing higher scores or new symptoms may prompt a closer look or a conversation about next steps. Entries in the "Other" and "Notes" sections can further open discussion about how symptoms are affecting daily life.

Bringing the tracker to appointments helps ensure everyone's on the same page, making it easier to recognize trends over time and consider timely adjustments when needed.

By combining patient-reported information with clinical judgment, care teams and patients can work together to keep GD1 management proactive and personalized.

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