At a Glance
Glycogen Storage Disease (GSD) Type Ia and Ib

Deficient Enzyme: GSD Ia: glucose-6-phosphatase; GSD Ib: glucose-6-phosphate translocase

Clinical presentation in undiagnosed patients or patients with poor metabolic control:[1]

Hypoglycemia, hepatomegaly, failure to thrive, short stature, lactic acidosis, hypertriglyceridemia, hyperuricemia, neutropenia (GSD Ib)

Treatment Goals:

- Prevent hypoglycemia
- Correct laboratory abnormalities
- Ensure adequate nutrient intake for age

Recommended Macronutrient Composition of Diet[1, 2, 3]

- Carbohydrate (CHO) (60-70% of energy intake)
  - Include calories provided by UCCS
  - Emphasize complex CHOs (limit to 15 g CHO per meal and 5g CHO per snack)
  - Limit fructose (2.5 g per meal)
  - Limit galactose and lactose (1 serving per day allowed)
  - Limit sucrose
  - Restrict simple sugar to <5 g per meal and 2-3 g per snack
- Protein (10-15% of energy intake)
  - Offer lean sources of protein
- Fat (<30% of energy intake for children older than 2 years)

Recommended Fasting Times[4]

Infants to 2 years old: 2 to 3.5 hours
Children and adults: 3 to 5 hours

Uncooked Cornstarch (UCCS) [2, 3]

- Initiating therapy
  - Begin UCCS at 9-12 months of age
  - Start with 1 g UCCS per dose, increasing by 1 g increments weekly as tolerated to goal
- For children <8 years old, calculate Bier Equation to determine dose of UCCS
  - Bier Equation
  - $Y = 0.0014X^3 - 0.214X^2 + 10.411X - 9.084$
  - Y = mg glucose per minute
  - X = weight in kg
  - i.e., X = 10 kg; Y = 75 mg/min (4.5 g/hr)

For individuals > 5 years old in USA (>2 years old in other countries), consider Glycosade®, a slow-release form of cornstarch that extends fasting time in some individuals[5]

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Enteral Formulas for GSD I*

***Ingredients may change, products should be reviewed before making a recommendation**

	Abbott abbottnutrition.com	Cambrooke cambrooke.com	Mead Johnson hcp.meadjohnson.com	Nestle nestlehealthscience.us	Nutricia nutriciametabolics.com
Infant (0-1 yr)	Elecare® Infant		Enfamil® ProSobee® Nutramigen® Pregestimil® PurAmino™	Alfamino® Infant	Neocate® Infant
Toddler & Young Children	Elecare Jr PediaSure® Peptide 1.0 unflavored PediaSure Peptide 1.5	Essential Care Jr.™ EquaCare Jr.®	Nutramigen Toddler PurAmino Jr	Alfamino Junior Peptamen Junior® Tolerex® Vivonex® Pediatric	Neocate Junior
Older Children & Adults	Ensure® Max Protein Osmolite®1.2 or 1.5			Isosource® HN Nutren® 1.5 or 2.0 Peptamen® 1.0 or 1.5 unflavored Vivonex Plus Vivonex TEN	

Supplementation

Sugar-free multivitamin/mineral, calcium, vitamin D_3 to meet Dietary Reference Intake for age and/or as indicated based off laboratory results

Consider probiotics

Monitoring

Nutrition:

Anthropometrics, dietary intake, physical findings

Laboratory:

Glucose, lactic acid, uric acid, triglycerides, cholesterol, liver function tests, markers for anemia (hemoglobin, hematocrit, MCV, ferritin, iron, folate, vitamin B_{12}), vitamin and mineral status (25-hydroxy vitamin D, zinc, trace minerals)

Glucose monitors and continuous glucose monitoring (CGM)

Point-of-care blood glucose testing (i.e. FreeStyle Lite) test prior to UCCS dosing

CGM (i.e. Dexcom) to assess trends over 24-hour period

References

1. Kishnani PS et al. Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics. Genet Med. 2014;16(11):e1.

2. Ross KM et al. Dietary Management of the Glycogen Storage Diseases: Evolution of Treatment and Ongoing Controversies. Adv Nutr. 2020;11(2):439-46.

3. Sowa, M. Nutritional Management of Glycogen Storage Diseases. In LE Bernstein, F Rohr, S van Calcar (Eds.) Nutritional Management of Inherited Metabolic Diseases (2nd Edition). Springer: 2021.

4. Weinstein DA et al. Inborn errors of metabolism with hypoglycemia: glycogen storage diseases and inherited disorders of gluconeogenesis. Pediatr Clin N Am. 2018;65(2):247-65

5.Weinstein DA et al. Short and long-term acceptability and efficacy of extended-release cornstarch in the hepatic glycogen storage diseases: results from the Glyde study. Orphanet J Rare Dis. 2024;19(1):258.