

**Reporting Title:** Uroporphyrinogen III Synthase, RBC  
**Performing Location:** Rochester

**Ordering Guidance:**  
This test is most appropriately used for pediatric patients.

This test measures uroporphyrinogen (UPG) III synthase to confirm congenital erythropoietic porphyria, which is typically seen in early infancy. It does not measure UPG I synthase (also known as porphobilinogen deaminase), the enzyme deficient in acute intermittent porphyria (AIP). For AIP (and UPG I synthase), order PBGD\_ / Porphobilinogen Deaminase, Whole Blood.

- Necessary Information:**
- 1. Include a list of medications the patient is currently taking.
  - 2. Date of transfusion, if performed

**Specimen Requirements:**  
All porphyrin tests on erythrocytes can be performed on one collection tube.

**Patient Preparation:** Patient should abstain from alcohol for 24 hours prior to specimen collection.

**Container/Tube:** Green top (sodium or lithium heparin)

**Specimen Volume:** 4 mL

**Collection Instructions:** Immediately place specimen on wet ice.

- Forms:**
- 1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:  
[-Informed Consent for Genetic Testing \(T576\)](#)  
[-Informed Consent for Genetic Testing-Spanish \(T826\)](#)
  - 2. If not ordering electronically, complete, print, and send a [Biochemical Genetics Test Request \(T798\)](#) with the specimen.

Specimen Type	Temperature	Time	Special Container
WB Heparin	Refrigerated	7 days	

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
80288	Uroporphyrinogen III Synthase, RBC	Numeric	RU	11066-8

LOINC® and CPT codes are provided by the performing laboratory.

**Supplemental Report:**  
No

CPT Code Information:

82657

Reference Values:

> or =75 Relative Units (normal)

See [The Heme Biosynthetic Pathway](#)