

## Galactose-1-Phosphate, Blood Spot

**Test ID: GAL1B**

### Useful for:

Monitoring dietary therapy of individuals with galactosemia due to deficiency of galactose-1-phosphate uridylyltransferase (GALT) or uridine diphosphate galactose-4-epimerase (GALE) in blood spots.

### Genetics Information:

Galactose-1-phosphate uridylyltransferase (GALT) deficiency is the most common cause of galactosemia and requires lifelong restriction of dietary galactose.

Galactose-1-phosphate is elevated in patients with galactosemia due to GALT deficiency or uridine diphosphate galactose-4-epimerase (GALE) deficiency, therefore is a suitable analyte for monitoring dietary compliance.

### Methods:

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

### Reference Values:

< or =1.0 mg/dL

### Specimen Requirements:

**Submit only 1 of the following specimen types:**

**Preferred Specimen Type:** Blood Spot

**Supplies:** Card-Blood Spot Collection (Filter Paper) (T493)

**Preferred:** Blood Spot Collection Card

**Acceptable:** Whatman Protein Saver 903 Paper, Revvity 226 filter paper, or blood collected in tubes containing EDTA and dried on filter paper

**Specimen Volume:** 2 Blood spots

**Collection Instructions:**

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. At least 2 spots should be complete (ie, unpunched).
3. Let blood dry on filter paper at ambient temperature in a horizontal position for

- a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
  5. Do not stack wet specimens.
  6. Keep specimen dry.

**Specimen Stability Information:**

Ambient (preferred) 91 days/Refrigerate 91 days/Frozen 91 days

**Acceptable Specimen Type:** Whole Blood

**Preferred:** Lavender top (EDTA)

**Acceptable:** Green top (NaHep) or Yellow top (ACD)

**Specimen Volume:** 2 mL

**Collection Instructions:** Send specimen in original tube. **Do not aliquot.**

**Specimen Stability Information:** Refrigerate (preferred) 48 hours/Frozen 14 days.

**Specimen Stability Information:**

Specimen Type	Temperature
Whole blood	Varies

**Cautions:**

Patients should wait 3 to 4 months after blood transfusion before blood collection for galactose-1-phosphate testing.

**CPT Code:**

84378

**Day(s) Performed:** Thursday

**Report Available:** 4 to 10 days

**Questions**

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.