

## Plasmalogens, Blood

**Test ID:** PGRBC

### Useful for:

Diagnosing patients with possible peroxisomal disorders, such as peroxisomal biogenesis disorders (Zellweger syndrome spectrum) and rhizomelic chondrodysplasia punctata (RCDP), including fatty acyl-CoA reductase 1 (FAR1) deficiency

Evaluating patients with abnormal newborn screen results for X-linked adrenoleukodystrophy who appear to have a different type of peroxisomal disorder, such as a Zellweger syndrome spectrum disorder.

An aid in the assessment of peroxisomal function

### Genetics Information:

This test measures plasmalogens and plasmalogen to fatty acid ratios for the purpose of diagnosis of peroxisomal biogenesis disorders (Zellweger syndrome spectrum) and rhizomelic chondrodysplasia punctata (RCDP), including fatty acyl-CoA reductase 1 (FAR1) deficiency.

### Methods:

Gas Chromatography-Mass Spectrometry (GC-MS)

### Reference Values:

HEXADECANAL-DIMETHYLACETAL, C16:0 DMA:  
> or =6.00 mcg/mL

OCTADECANAL-DIMETHYLACETAL, C18:0 DMA:  
> or =9.00 mcg/mL

9Z-OCTADECENAL-DIME ACETAL C18:1DMA:  
> or =2.00 mcg/mL

C16:0 DMA/C16:0:  
> or =0.018

C18:0 DMA/C18:0:  
> or =0.040

## Specimen Requirements:

<b>Patient Preparation:</b>	Collect either prior to or 6 weeks after a blood transfusion
<b>Preferred:</b>	Lavender top (EDTA)
<b>Acceptable:</b>	Green top (sodium or lithium heparin), yellow top (ACD solution A or ACD solution B)
<b>Specimen Volume:</b>	2 mL
<b>Collection Instructions:</b>	Send specimen in original tube.
<b>Minimum Volume:</b>	0.5 mL

## Specimen Stability Information:

Specimen Type	Temperature	Time
Whole Blood	Refrigerate (preferred)	14 days
	Ambient	14 days

## Cautions:

The results of testing performed in erythrocytes are invalid following a transfusion; therefore, collect specimen either prior to or 6 weeks after blood transfusion.

## CPT Code:

82542

**Day(s) Setup:** Wednesday; 8 a.m.

**Analytic Time:** 3 days

## Questions

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