
Plasmalogens, Blood Spot

Test ID: PGDBS

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 =7.00 mcg/mL

OCTADECANAL-DIMETHYLACETAL, C18:0 DMA:
> or =12.00 mcg/mL

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

C16:0 DMA/C16:0:
> or =0.012

C18:0 DMA/C18:0:
> or =0.050

Specimen Requirements:

Specimen must be collected either prior to or 6 weeks after a blood transfusion.

Submit only 1 of the following specimens:

Preferred:

Specimen Type: Blood spot

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

Preferred: Blood Spot Collection card

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper, Munktell filter paper, Whatman Protein Saver 903 paper, or blood collected in tubes containing ACD, EDTA, or heparin spotted and dried on filter paper

Specimen Volume: 2 blood spots

Collection Instructions:

1. Let blood dry completely on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
2. At least 2 spots should be completed (ie, unpunched)
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information:

Ambient (preferred)	90 days
Refrigerated	90 days
Frozen	90 days

Minimum Volume: 1 blood spot

Acceptable:

Specimen Type: Whole Blood

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium or lithium heparin), yellow top (ACD A or ACD B)

Specimen Volume: 2 mL

Collection Instructions: Send specimen in original tube.

Specimen Stability Information:

Refrigerate (preferred)	14 days
Ambient	11 days

Specimen Stability Information:

Specimen Type	Temperature
Whole Blood	Varies

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.