
Reporting Title: Plasmalogens, BS**Performing Location:** Rochester**Additional Testing Requirements:**

If peroxisomal biogenesis disorders (Zellweger syndrome spectrum) are suspected, also order very long chain fatty acids (POX / Fatty Acid Profile, Peroxisomal [C22-C26], Serum; or POXP / Fatty Acid Profile, Peroxisomal [C22-C26], Plasma), bile acids (BAIPD / Bile Acids for Peroxisomal Disorders, Serum), and pipecolic acid (PIPU / Pipecolic Acid, Random, Urine).

If rhizomelic chondrodysplasia punctata (RCDP) is suspected, also order very long chain fatty acids (POX / Fatty Acid Profile, Peroxisomal [C22-C26], Serum), which includes phytanic and pristanic acid analysis.

Necessary Information:

1. Reason for testing is required
2. Date of blood transfusion, if performed.
3. Biochemical Genetics Patient Information (T602) is recommended, but not required, to be filled out and sent with the specimen to aid in the interpretation of test results.

Specimen Requirements:

Submit only 1 of the following specimens:

Preferred:

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

Specimen Type: Blood spot

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

Container/Tube:

Preferred: Blood spot collection card

Acceptable: Local newborn screening card, PerkinElmer 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. 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. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
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 90days/Frozen 90 days

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Acceptable

Specimen Type: Whole Blood

Container/Tube:

Preferred: Lavender top (EDTA)

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

Specimen Volume: 2 mL

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

Specimen Stability Information: Refrigerate (preferred) 14 days/Ambient 11 days

Specimen Minimum Volume:

Blood spot: 1

Whole blood: 0.5 mL

Forms:

1. Biochemical Genetics Patient Information (T602)

2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
PGDBS	BG724	Reason for Referral: <ul style="list-style-type: none">• Follow up abnormal NBS for C26 LPC• MRI findings• Molecular findings• Skeletal abnormalities• Treatment monitoring• Not provided	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
609665	Hexadecanal-Dimethylacetal, C16 DMA	Numeric	mcg/mL	In Process
609666	Octadecanal-Dimethylacetal, C18 DMA	Numeric	mcg/mL	In Process
609667	9Z-Octadecenal-DiMe acetal C18:1DMA	Numeric	mcg/mL	In Process
609670	C16 DMA/C16:0	Alphanumeric		In Process

Result ID	Reporting Name	Type	Unit	LOINC®
609671	C18 DMA/C18:0	Alphanumeric		In Process
BG724	Reason for Referral	Alphanumeric		42349-1
609673	Reviewed By	Alphanumeric		18771-6
609674	Interpretation	Alphanumeric		59462-2

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

Hexadecanal-Dimethylacetal, C16:0 DMA
> or =7.00 mcg/mL

Octadecanal-Dimethylacetal, C18:0 DMA
> or =12.00 mcg/mL

9Z-Octadecenal-Dimethylacetal C18:1DMA
> or =2.00 mcg/mL

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

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