

Test Definition: PGRBC

Plasmalogens, Blood

Reporting Title: Plasmalogens, RBC

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.

Shipping Instructions:

Whole blood should be sent refrigerated.

Necessary Information:

- 1. Reason for testing is required
- 2. Date of blood transfusion, if performed.
- 3. <u>Biochemical Genetics Patient Information</u> (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:

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

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: 5 mL

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

Forms:

- 1. Biochemical Genetics Patient Information (T602) (recommended, but not required)
- 2. If not ordering electronically, complete, print, and send a <u>Biochemical Genetics Test Request</u> (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	14 days	
	Ambient	14 days	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
PGRBC	BG726	Reason for Referral:	Answer List	Yes
		Follow up abnormal NBS for C26 LPC		
		MRI findings		



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	Molecular findings	
	Skeletal abnormalitiesTreatment monitoring	
	Not provided	

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
609676	Hexadecanal-Dimethylacetal, C16 DMA	Numeric	mcg/mL	In Process
609677	Octadecanal-Dimethylacetal, C18 DMA	Numeric	mcg/mL	In Process
609678	9Z-Octadecenal-DiMe acetal C18:1DMA	Numeric	mcg/mL	In Process
609681	C16 DMA/C16:0	Alphanumeric		In Process
609682	C18 DMA/C18:0	Alphanumeric		In Process
BG726	Reason for Referral	Alphanumeric		42349-1
609684	Reviewed By	Alphanumeric		18771-6
609685	Interpretation	Alphanumeric		59462-2

 $\ensuremath{\mathsf{LOINC}}\xspace^{\ensuremath{\texttt{@}}}$ and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

Hexadecanal-Dimethylacetal, C16:0 DMA:

> or =6.00 mcg/mL

Octadecanal-Dimethylacetal, C18:0 DMA:

> or =9.00 mcg/mL

9Z-Octadecenal-DiMethylacetal C18:1 DMA:

> or =2.00 mcg/mL

C16:0 DMA/C16:0:

> or =0.018

C18:0 DMA/C18:0:

> or =0.040