Peroxisomal Disorders

High Precision and Specificity Testing Improves Disease Detection

Markedly reduced plasmalogen levels are a hallmark of impaired peroxisome biogenesis, as seen in patients with Zellweger spectrum disorders (ZSDs). Plasmalogen deficiency is also the main biochemical finding in rhizomelic chondrodysplasia punctata (RCDP) and correlates with disease severity. Determining plasmalogen levels aids the diagnosis of ZSDs and RCDP and is critical for evaluating patients with developmental delays, seizures, hypotonia, congenital abnormalities, skeletal defects, and liver disease. It can also be useful in evaluating infants who screen positive for the peroxisomal disorder X-linked adrenoleukodystrophy but may have a ZSD.

ACCESS TESTING WITH GREATER PRECISION AND SPECIFICITY:

DETECTING PLASMALOGEN DEFICIENCY:

- Is essential for the diagnosis of peroxisome biogenesis defects, particularly RCDP
- Supports the results of molecular genetic testing by providing functional evidence
- Provides further information on disease severity and progression
- Offers a quantitative biomarker to monitor targeted therapies (e.g., plasmalogen augmentation)

AVAILABLE TEST

3019936	**Plasmalogens (Red Blood Cells)** Use for the diagnostic evaluation of patients with developmental delays, neurological symptoms, hypotonia, liver dysfunction, or suspected metabolic disorders.

WHY CHOOSE ARUP

High precision and specificity: Quantitative liquid chromatography-tandem mass spectrometry (LC-MS/MS) provides plasmalogen quantitation as absolute concentrations.

Age-specific reference intervals: Established normal ranges from birth to adulthood improve diagnostic accuracy across all ages.

Effective differentiation of disorders: ARUP's assay clearly distinguishes patients with RCDP and identifies severe versus mild phenotypes.

ARUP LABORATORIES

aruplab.com/plasmalogen