

Aminoacylase-1 Deficiency, Urine

Test ID: AACYL

Useful for:

Follow-up quantitation of abnormal organic acid elevations of N-acetylated amino acids, in particular N-acetylalanine, N-acetylglycine, N-acetylmethionine, and N-acetylglutamic acid

Diagnosis of individuals with aminoacylase-1 deficiency

Evaluating patients with neurologic and psychiatric symptoms of unknown etiology

Methods:

Gas Chromatography Mass Spectrometry (GC-MS)

Reference Values:

N-acetylglycine: < or =15.00 mmol/mol creatinine

N-acetyl alanine: < or =10.00 mmol/mol creatinine

N-acetylglutamic acid: < or =20.00 mmol/mol creatinine

N-acetylmethionine: < or =5.00 mmol/mol creatinine

Necessary Information:

1. Patient's age and sex are required.
2. Include family history, clinical condition (asymptomatic or acute episode), diet, and drug therapy information

Specimen Requirements:

Supplies: Urine Tubes, 10 mL (T068)

Container/Tube: Plastic, 10-mL urine tube

Specimen Volume: 10 mL

Pediatric: If insufficient collection volume, submit as much specimen as possible in a single container; the laboratory will determine if volume is sufficient for testing.

Collection Instructions: 1. Collect a random urine specimen (first morning void preferred).
2. No preservative.

Minimum Volume: 3 mL

Specimen Stability Information:

Specimen Type	Temperature	Time
Urine	Frozen (preferred)	90 days
	Refrigerated	60 days

Cautions:

No significant cautionary statements

CPT Code:

82542

Day(s) Performed: Monday, Wednesday, Friday **Report Available:** 3-6 days

Questions

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