

Adenosine Monophosphate Deaminase 1 (AMPD1) Deficiency: *AMPD1* Two Mutation Panel

Test Code: PAMPD

Turnaround time: 2 weeks

CPT Codes: 81479 x1

Condition Description

Adenosine monophosphate deaminase 1 (AMPD1) deficiency, also known as myoadenylate deaminase (MADA) deficiency, is a disorder of purine metabolism that leads to a deficiency in the production of ATP. It is the most common enzyme deficiency identified in muscle, with a prevalence of almost 2% in the general population. The typical age of presentation is late adolescence to early adulthood. Affected individuals have generalized exertional muscle pain, cramps and fatigue. Other presenting features include post-exertional myoglobinuria and rhabdomyolysis. Completely asymptomatic individuals have also been reported.

Serum creatine kinase (CK) is usually normal or only slightly elevated. Aerobic exercise testing is typically normal. Muscle histology is normal but muscle histochemistry shows reduced AMPD1 enzyme activity. AMPD1 deficiency is caused by mutations in the *AMPD1* gene (1p21). AMPD1 deficiency is an autosomal recessive condition.

Two mutations, c.133C>T (p.Q45X, previously known as p.Q12X) and c.242C>T (p.P81L, previously known as p.P48L), account for the majority of reported mutations in Caucasians and African Americans. Full gene sequence analysis is also available for individuals with documented AMPD1 deficiency when no or one mutation identified by common mutation testing.

[Click here](#) for the OMIM summary on this condition.

Reference:

- Brian A. van Adel, et al. Metabolic Myopathies: Update 2009. J Clin Neuromusc Dis. 2009, 10(3): 97-121.

Genes

[AMPD1](#)

Indications

This test is indicated for:

- Confirmation of a biochemical or clinical diagnosis of AMPD1 deficiency
- Carrier testing in adults with a family history of AMPD1 deficiency

Methodology

Presence/absence of the p.Q12X and p.P48L mutations are detected by PCR amplification and sequencing of the resulting fragments.

Detection

All p.Q12X and p.P48L mutant alleles will be detected by this assay. Some studies have found that these two mutations account for the majority of reported mutations in Caucasians and African Americans. Prevalence of *AMPD1* mutations in other ethnic groups is currently unknown.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection Kit

Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:

Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

Type: DNA, Isolated

Specimen Requirements:

Microtainer

8µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/μL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)**Specimen Requirements:**

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Special Instructions

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Related Tests

- Full gene sequence analysis of the *AMPD1* gene is available if common mutation testing does not identify two mutations in a clinically and biochemically affected individual.
- A two-tiered rhabdomyolysis panel that includes testing for the two common *AMPD1* mutations is also available.
- [Custom diagnostic mutation analysis \(KM\)](#) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.