Adenosine Monophosphate Deaminase (AMPD) Deficiency, Erythrocytic: \textit{AMPD3} Gene Deletion/Duplication

\textbf{Test Code: DAMP3}
\textbf{Turnaround time: 2 weeks}
\textbf{CPT Codes: 81228 x1}

\section*{Condition Description}

Adenosine monophosphate deaminase (AMPD) is found in muscle, liver, and erythrocytes. The \textit{AMPD3} gene (11p15.4) encodes the AMPD found in erythrocytes. Individuals with two mutations in their \textit{AMPD3} gene are healthy and have no hematologic disorders. Their ATP levels were approximately 150\% higher in the AMPD deficient red cells when compared with control cells. Additionally, degradation of the adenine nucleotide was slower in the deficient red cells when compared with control cells. AMPD deficiency is inherited in an autosomal recessive pattern.

For patients with suspected AMPD deficiency, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

\section*{References:}

- OMIM \#612874: AMPD deficiency, erythrocytic
- OMIM \#102772: \textit{AMPD3} gene

\section*{Genes}

\textbf{AMPD3}

\section*{Indications}

This test is indicated for:

- Confirmation of a clinical diagnosis of adenosine monophosphate deaminase deficiency, erythrocytic in an individual in whom sequence analysis was negative.
- Carrier testing in adults with a family history of adenosine monophosphate deaminase deficiency, erythrocytic in whom sequence analysis was negative.

\section*{Methodology}

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

\section*{Detection}

Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

\section*{Specimen Requirements}

Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

\textbf{Type: Whole Blood}

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

\textbf{Type: Saliva}

Specimen Requirements:
Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

### Special Instructions

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

### Related Tests

- Sequence analysis of the *AMPD3* gene is available and is required before deletion/duplication analysis.
- Sequence and deletion/duplication analysis for the *AMPD1* gene are 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 only for known familial mutations to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.