

Overview

Useful For

Assessment of adenylate kinase activity as part of the evaluation of chronic nonspherocytic hemolytic anemia

Method Name

Only available as part of a profile. For more information see:

-HAEV1 / Hemolytic Anemia Evaluation, Blood

-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Kinetic Spectrophotometry

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

Only available as part of a profile. For more information see:

-HAEV1 / Hemolytic Anemia Evaluation, Blood

-EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood

Reject Due To

Gross hemolysis Reject

Specimen Minimum Volume

1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated (preferred)	20 days	

Clinical & Interpretive

Clinical Information

Adenylate kinase (AK) is a monomeric enzyme that catalyzes the nucleotide phosphoryl interconversion of adenosine triphosphate (ATP) and adenosine monophosphate (AMP) to 2 molecules of adenosine diphosphate (ADP). The level of enzyme activity in neonates is normally mildly to moderately lower than in adults. AK deficiency (OMIM 612631) is a rare cause of autosomal recessive nonspherocytic hemolytic anemia.

Although rare, AK deficient-associated anemia has been described in multiple families of varied ethnic origin. Those individuals with heterozygous genetic alterations are predominantly asymptomatic and show a normal phenotype. Those individuals with homozygous or compound heterozygous genetic alterations display congenital chronic nonspherocytic hemolytic anemia (hemoglobin [Hb] levels of 8-9 g/dL) with hyperbilirubinemia and gallstones. Patients typically present at birth or in early childhood. Some patients have psychomotor impairment, although the pathogenesis is not well understood. Concurrent glucose 6-phosphate dehydrogenase (G6PD) deficiency exacerbates the anemia (Hb 6 g/dL). AK activity levels range from 0% to 44%, although most show less than 30% activity. Carriers have normal to only mildly decreased enzyme activity (1). Patients may respond well to splenectomy.

Reference Values

Only available as part of a profile. For more information see:

- HAEV1 / Hemolytic Anemia Evaluation, Blood
- EEEV1 / Red Blood Cell (RBC) Enzyme Evaluation, Blood
- > or =12 months: 195-276 U/g Hb

Reference values have not been established for patients who are less than 12 months of age.

Interpretation

In adenylate kinase deficiency, values are expected to be less than 30% of normal mean, although this value should be interpreted in the context of age of the patient and other enzyme values.

Cautions

Recent transfusion may mask the patient's intrinsic enzyme activity and cause unreliable results.

Adenylate kinase (AK) enzyme activity can normally be mildly to moderately decreased in neonates.

Clinical Reference

1. Niizuma H, Kanno H, Sato A, Ogura H, Imaizumi M: Splenectomy resolves hemolytic anemia caused by adenylate kinase deficiency. *Pediatr Int.* 2017 Feb;59(2):228-230
2. Rapley S, Harris H: Red cell adenylate kinase activity in AK1 and AK 2-1 phenotypes. *Annals of Human Genetics.* 1970;33:361-364. doi: 10.1111/j.1469-1809
3. Mohrenweiser HW. Frequency of enzyme deficiency variants in erythrocytes of newborn infants. *Proc Natl Acad Sci U S A.* 1981 Aug;78(8):5046-5050
4. Corrons JL, Garcia E, Tusell JJ, Varughese KI, West C, Beutler E: Red cell adenylate kinase deficiency: molecular study of 3 new mutations (118G>A, 190G>A, and GAC deletion) associated with hereditary nonspherocytic hemolytic anemia. *Blood.* 2003 Jul 1;102(1):353-356
5. Toren A., Brok-Simoni F, Ben-Bassat I, et al: Congenital haemolytic anaemia associated with adenylate kinase deficiency. *Brit. J. Haemat.* 1994;87:376-380
6. Bianchi P, Zappa M, Bredi E, et al: A case of complete adenylate kinase deficiency due to a nonsense mutation in AK-1 gene (arg107-to-stop, CGA-to-TGA) associated with chronic haemolytic anaemia. *Brit. J. Haemat.* 1999;105:75-79
7. Lachant NA, Zerez CR, Barredo J, et al: Hereditary erythrocyte adenylate kinase deficiency: A defect of multiple phosphotransferases? *Blood.* 1991;77(12):2774-2784
8. Koralkova P, van Solinge WW, van Wijk R: Rare hereditary red blood cell enzymopathies associated with hemolytic anemia-pathophysiology, clinical aspects and laboratory diagnosis. *Int J Lab Hematol.* 2014;36:388-397

Performance

Method Description

Adenylate kinase (myokinase) catalyzes the dismutation of adenosine diphosphate (ADP) into adenosine monophosphate and adenosine triphosphate. In this assay, the reverse reaction is measured by following the formation of ADP with pyruvate kinase and lactate dehydrogenase reactions resulting in 1,4-dihyronicotinamide adenine dinucleotide (NADH) being oxidized to NAD(+). The decrease in absorbance that occurs as NADH is oxidized is measured spectrophotometrically at 340 nm by an automated chemistry analyzer. (Beutler E: Red Cell Metabolism. A Manual of Biochemical Methods. 3rd ed. Grune and Stratton; 1984:93-95; van Solinge WW, van Wijk: Enzymes of the red blood cell. In: Rifai N, Horvath AR, Wittwer CT: eds. Tietz Textbook of Clinical Chemistry and Molecular Diagnostics. 6th ed. Elsevier; 2018:chap 30)

PDF Report

No

Specimen Retention Time

7 days

Performing Laboratory Location

Rochester

Fees & Codes**Test Classification**

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82657