MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: August 19, 2022
Effective Date: September 6, 2022

KLF1 Full Gene Sequencing, Varies

Test ID: KLF1

Useful for:

This test is intended to aid in the diagnosis and carrier detection of KLF1 sequence alterations that are reported to be responsible for increased fetal hemoglobin (HbF) and hemoglobin A2 (HbA2). Variants in KLF1 have also been associated with severe neonatal anemia and congenital dyserythropoietic anemia.

Methods:

Polymerase Chain Reaction (PCR) Amplification followed by Sanger Sequencing

Reference Values:

An interpretive report will be provided

Specimen Requirements:

Submit only 1 of the following specimens:

Preferred:
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD) or green top (heparin)
Specimen Volume: 4 mL
Minimum Volume: 1 ml
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
3. Label specimen as blood.
Specimen Stability Information: Ambient 14 days (preferred)/ Refrigerated 30 days

Acceptable:
Specimen Type: Extracted DNA from whole blood
Container/Tube: 1.5- to 2-mL tube with indication of volume and concentration of DNA
Specimen Volume: Entire specimen
Minimum Volume: 50 mcL at 50 ng/mcL concentration
Collection Instructions: Label specimen as extracted DNA from blood and provide indication of volume and concentration of the DNA
Specimen Stability Information: Frozen/Refrigerate/Ambient

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Varies	

Cautions:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Individuals may have a variant, deletion, or duplication in the gene tested that is not identifiable by the described testing methodology. Rare variants (polymorphisms) exist and could lead to false negative results. In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test. This assay will not detect deep intronic or large deletion-insertion (delins) sequence alterations.

Rare, undocumented variants (ie, polymorphisms) under the primers can cause polymerase chain reaction failure.

Patients who have received an allogenic blood transfusion within the preceding 6 weeks, or who have received an allogenic blood or marrow transplant can have inaccurate genetic test results due to presence of donor DNA.

CPT Code:
81479

Day(s) Performed: Monday through Friday
Report Available: 28 to 42 days

Questions
Contact Connie Penz, Laboratory Technologist Resource Coordinator at 800-533-1710.