MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: November 19, 2024
Effective Date: December 19, 2024

Alpha Globin Cluster Locus Deletion/Duplication, Varies

Test ID: AGDD

Useful for:

- Diagnosis of alpha-thalassemia
- Prenatal diagnosis of deletional alpha-thalassemia
- Carrier screening for individuals from high-risk populations for alpha-thalassemia
- This test is **not useful for** diagnosis or confirmation of beta-thalassemia or hemoglobinopathies.

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No

Testing Algorithm:

- For prenatal specimens only: If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture will be added at an additional charge.
- For any prenatal specimen that is received, maternal cell contamination studies will be performed at an additional charge. A maternal whole blood specimen is required to perform this test.

Ordering Guidance:

Sequence variants, other than the alpha T-Saudi and hemoglobin constant spring alterations, are not detected by this assay. For detection of single point and other nondeletion variants, order WASEQ / Alpha Globin Gene Sequencing, Varies, if clinically indicated.

Methods:

Dosage Analysis by Polymerase Chain Reaction (PCR)/Multiplex Ligation-Dependent Probe Amplification (MLPA)

Reference Values:

An interpretive report will be provided.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type:	**Whole blood**
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any other anticoagulant tube
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. **Do not aliquot**.
Specimen Stability Information:	Ambient (preferred) 4 days/Refrigerated 14 days
Minimum Volume:	1 mL

Specimen Type:	**Saliva**
Patient Preparation:	Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies:	Saliva Swab Collection Kit (T786)
Specimen Volume:	1 Swab
Collection Instructions:	Collect and send specimen per kit instructions.
Specimen Stability Information:	Ambient 30 days
Additional Information:	Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Prenatal Specimens: Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type:	**Amniotic fluid**
Container/Tube:	Amniotic fluid container
Specimen Volume:	20 mL
Specimen Stability Information:	Refrigerated (preferred)/Ambient
Additional Information:	1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur. **2. All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type:	**Confluent cultured cells**
Container/Tube:	T-25 flask
Specimen Volume:	2 Flasks

Collection Instructions:	Submit confluent cultured cells from another laboratory.
Specimen Stability Information:	Ambient (preferred)/Refrigerated
Additional Information:	**All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Additional Testing Requirements:

All prenatal specimens must be accompanied by a maternal or gestational carrier blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Cautions:

- Hemoglobin electrophoresis should usually be done prior to this test to exclude other diagnoses.
- In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.
- Rare alterations (ie, polymorphisms) exist that could lead to false-negative or false-positive results. If the results obtained do not match the clinical findings, additional testing should be considered.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given is inaccurate or incomplete.

CPT Code:

81269
88235-Tissue culture for amniotic fluid (if appropriate)
88240-Cryopreservation (if appropriate)
81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

Day(s) Performed: Monday, Wednesday **Report Available:** 9 to 13 days

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.