



# Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome (aHUS)/Thrombotic Microangiopathy (TMA) /Complement 3 Glomerulopathy (C3G) Gene Panel, Varies

## Overview

### Useful For

Providing a genetic evaluation for patients with a personal or family history suggestive of atypical hemolytic uremic syndrome (aHUS), thrombotic microangiopathy (TMA), or complement 3 glomerulopathy (C3G)

Establishing a diagnosis of genetic aHUS, TMA, or C3G and, in some cases, allowing for appropriate management and surveillance for disease features based on the gene involved

Identifying variants in genes encoding complement alternate pathway components and specific coagulation pathway genes known to be associated with increased risk for aHUS, TMA, and C3G allowing for predictive testing of at-risk family members

Providing genetic information that may be considered when making treatment decisions, including duration of therapy and recurrence risk, as well as consideration of transplantation

### Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No

### Genetics Test Information

This test utilizes next-generation sequencing to detect single nucleotide, small deletion-insertion, and copy number variants in 15 genes associated with atypical hemolytic uremic syndrome (aHUS), thrombotic microangiopathy (TMA), and complement 3 glomerulopathy (C3G): *ADAMTS13*, *C3*, *C5*[Chr9(GRCh37):g.123759950-123759973 only], *CD46* (*MCP*), *CFB*, *CFH*, *CFHR1*, *CFHR2*, *CFHR3*, *CFHR4*, *CFHR5*, *CFI*, *DGKE*, *MMACHC*, and *THBD*. See [Targeted Genes and Methodology Details for Atypical Hemolytic Uremic Syndrome / Thrombotic Microangiopathy / Complement 3 Glomerulopathy Gene Panel](#) in Method Description for additional details.

Identification of a pathogenic variant may assist with diagnosis, prognosis, clinical management, familial screening, and

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

genetic counseling for aHUS, TMA, and C3G.

### Testing Algorithm

#### Prenatal specimens:

If an amniotic fluid specimen or cultured amniocytes are received, an amniotic fluid culture will be performed at an additional charge.

If a chorionic villi specimen or cultured chorionic villi are received, a fibroblast culture will be performed at an additional charge.

For any prenatal specimen that is received, maternal cell contamination testing will be performed at an additional charge.

#### Skin biopsy or cultured fibroblast specimens:

For skin biopsy or cultured fibroblast specimens, a fibroblast culture will be performed at an additional charge. If viable cells are not obtained, the client will be notified.

#### Cord blood:

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

### Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Hereditary Renal Genetic Testing Patient Information](#)
- [Targeted Genes and Methodology Details for Atypical Hemolytic Uremic Syndrome / Thrombotic Microangiopathy / Complement 3 Glomerulopathy Gene Panel](#)

### Method Name

Sequence Capture and Amplicon-Based Next-Generation Sequencing (NGS)

### NY State Available

Yes

## Specimen

### Specimen Type

Varies

### Ordering Guidance

Due to atypical hemolytic uremic syndrome genotype-phenotype complexity, targeted testing for familial variants will not be accepted without approval from the laboratory; call 800-533-1710 to discuss testing options with a genetic

# Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

counselor.

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information, see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies. To modify this panel via CGPH, use the Hereditary Renal Conditions disease state for step 1 on the [Custom Gene Ordering Tool](#).

## Specimen Required

**Patient Preparation:** A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

## Submit only 1 of the following specimens:

**Specimen Type:** Whole blood

**Container/Tube:** Lavender top (EDTA) or yellow top (ACD)

**Specimen Volume:** 3 mL

### Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**
3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information

**Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

### Additional Information:

1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

**Specimen Type:** Skin biopsy

**Supplies:** Fibroblast Biopsy Transport Media (T115)

**Container/Tube:** Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

**Specimen Volume:** 4-mm Punch

**Specimen Stability Information:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:** Specimen will only be tested after culture.

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

**Specimen Type:** Cultured fibroblasts

**Source:** Skin or tissue

**Container/Tube:** T-25 flask

**Specimen Volume:** 2 Flasks

**Collection Instructions:** Submit confluent cultured fibroblast cells from a skin or tissue biopsy.

**Specimen Stability Information:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:**

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

**Specimen Type:** Extracted DNA

**Container/Tube:**

**Preferred:** Screw Cap Micro Tube, 2 mL with skirted conical base

**Acceptable:** Matrix tube, 1 mL

**Collection Instructions:**

1. The preferred volume is at least 100 µL at a concentration of 75 ng/µL.
2. Include concentration and volume on tube.

**Specimen Stability Information:** Frozen (preferred) 1 year/Ambient/Refrigerated

**Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

### 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:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:** Specimen will only be tested after culture.

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks are required to culture amniotic fluid before genetic testing can occur.
3. **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

**Specimen Type:** Confluent cultured amniocytes

This does not include cultured chorionic villi.

**Container/Tube:** T-25 flask

**Specimen Volume:** 2 Full flasks

**Collection Instructions:** Submit confluent cultured amniocytes from another laboratory.

**Specimen Stability Information:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:**

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing.
3. **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:** Chorionic villi

**Container/Tube:** 15-mL tube containing 15 mL of transport media

**Specimen Volume:** 20 mg

**Specimen Stability Information:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:** Specimen will only be tested after culture.

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.
3. **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:** Cultured chorionic villi

**Container/Tube:** T-25 flasks

**Specimen Volume:** 2 Full flasks

**Collection Instructions:** Submit confluent cultured cells from another laboratory.

**Specimen Stability Information:** Ambient (preferred) <24 hours/Refrigerated <24 hours

**Additional Information:**

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing.
3. **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

### Forms

**1. New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:

[-Informed Consent for Genetic Testing \(T576\)](#)

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

[-Informed Consent for Genetic Testing-Spanish \(T826\)](#)

2. [Hereditary Renal Genetic Testing Patient Information \(T918\)](#)

3. [If not ordering electronically, complete, print, and send a Renal Diagnostics Test Request \(T830\)](#) with the specimen.

### Specimen Minimum Volume

See Specimen Required

### Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

### Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

### Clinical & Interpretive

#### Clinical Information

Thrombotic microangiopathy (TMA) is a pathologic condition characterized by abnormalities in the walls of small blood vessels (arterioles and capillaries) that result in microvascular thrombosis. Typically, they feature microangiopathic hemolytic anemia (MAHA) and thrombocytopenia, but these features may not be apparent in kidney-limited disease. Laboratory findings may include anemia, thrombocytopenia, presence of schistocytes on peripheral smear, elevated lactate dehydrogenase, and elevated serum creatinine.(1,2) The main categories of TMA include complement-mediated TMA (CM-TMA; also known as atypical hemolytic syndrome [aHUS]), thrombotic thrombocytopenic purpura (TTP), Shiga toxin-mediated hemolytic uremic syndrome, and drug-induced TMA. Due to the overlapping clinical features, laboratory testing is useful in differentiated these disorders.(3)

Complement-mediated thrombotic microangiopathy (CM-TMA) (aHUS) is a well-recognized disease entity characterized by complement activation in the microvasculature. Abnormalities of the alternate pathway of complement, which may be inherited (genetic) or acquired, underlie both the sporadic and familial forms of the disease and are identified estimated to occur in approximately 60% of affected individuals.(3,4) Unlike many other monogenic disorders of the immune system, multiple hits may be required for disease manifestation, which may include a trigger event (transplantation, pregnancy, malignant hypertension, autoimmune disorders, sepsis, malignancy, etc) and one or more contributing genetic variants or risk haplotypes in the alternate pathway complement genes.(3) Individuals with genetic CM-TMA (aHUS) may experience relapse even after complete recovery following the presenting episode.

Thrombotic thrombocytopenic purpura is a rare clinical entity but is important to diagnose properly since it is associated with very high mortality (90%) if untreated. Mortality can be reduced by early plasma exchange. Congenital TTP is due to genetic defects in the ADAMTS13 gene, while acquired TTP is related to autoantibodies against ADAMTS13, which reduces function. While TTP was initially characterized by thrombocytopenia, MAHA, fluctuating neurological signs,

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

kidney failure and fever, not all of these features may be present in the manifestation of the disease.(1,2)

The hereditary form of CM-TMA is characterized by the presence of disease-causing variants in one or more of the genes known to be associated with aHUS, irrespective of familial history, or when two or more members of the same family are affected by the disease at least 6 months apart and exposure to a common triggering infectious agent has been excluded.(3) A patient may have both genetic variants in the alternative complement pathway and autoantibodies. While genetic testing may be used during the diagnostic work-up, the presence of disease-causing variants may also alter recurrence risk and impact decisions related to continuation of anti-complement therapy after resolution of symptoms.

Complement 3 glomerulopathies (C3G) include dense deposit disease and C3 glomerulonephritis and are characterized by C3 deposition within the glomeruli. In these disorders, the activity of the C3 convertase is increased by C3 nephritic factors, which are antibodies that stabilize the convertase, or loss of complement regulator activity, which may be due to genetic variants, autoantibodies, or other immunoglobulins. C3G may be preceded by an upper respiratory tract infection in some cases. Patients typically have proteinuria or hematuria and may present with variable kidney impairment. In addition to medical therapy, patients may be treated with kidney transplantation; however, disease recurrence and graft loss may occur.(4)

It is important to note that while TMA and C3G are associated with complement dysregulation, disease-causing variants in these genes may also result in complement deficiency, which is associated with recurrent infections with encapsulated pathogens or connective tissue diseases with no evidence of aHUS/TMA.(5)

Two risk alleles associated with increased susceptibility to aHUS/TMA and variants in C5 associated with poor response to anticomplement therapy are also included on this panel to aid in risk assessment:

-CFH-H3 risk haplotype: The variants that comprise this risk haplotype are common in the general population, but in the context of additional pathogenic genetic and environmental factors, the presence of this risk haplotype is associated with an increased risk for development or progression of atypical hemolytic uremic syndrome.(6)

-MCP/CD46 risk haplotype: The variants that comprise this risk haplotype are common in the general population, but in the context of additional pathogenic genetic and environmental factors, the presence of this risk haplotype is associated with an increased risk for development or progression of atypical hemolytic uremic syndrome.(6)

-C5 genotype: Two variants, p.Arg885His and p.Arg885Cys, have been associated with poor response to eculizumab.(7)

In addition, assessment of the CFHR gene cluster (CFHR1, CFHR2, CFHR3, CFHR4, and CFHR5) is included. Homozygous deletion of CFHR1 is enriched in patients with complement factor H (FH) autoantibodies (79%-89%), suggesting homozygous CFHR1 deletions may be a risk factor for FH autoantibody development.(8) However, variation, including simple variants and copy number variants, at this locus is common in the general population and at this time the clinical significance of other variants within this gene cluster is uncertain.

### Reference Values

An interpretive report will be provided.

### Interpretation

# Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

---

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.<sup>(9)</sup> Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

## Cautions

### Clinical Correlations:

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of at-risk individuals.

To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact the Mayo Clinic Laboratories genetic counselors at 800-533-1710.

### Technical Limitations:

Next-generation sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions; assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

This analysis targets single and multi-exon deletions/duplications; however, in some instances single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions), including hybrid alleles formed between *CFH* and *CFHR* genes, may not be detected.

Deletion/duplication events that extend past the genes included on the panel may occur. In these instances, genes included in the ordered test are provided on the report and interpreted, and genomic breakpoints are reported if they are confirmed. However, copy number variants for genes not listed in the Method Description are typically not reported or interpreted for haploinsufficiency/triplosensitivity. CMACB / Chromosomal Microarray, Congenital, Blood; WESPR / Panel to Whole Exome Sequencing Reflex Test, Varies; or WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies is recommended for a full interpretation of deletions/duplications predicted to extend past the genes included on

---

the panel.

This test is not designed to detect low levels of mosaicism or to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Genes may be added or removed based on updated clinical relevance. For detailed information regarding gene specific performance and technical limitations, see Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent non-leukocyte reduced blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

#### Reclassification of Variants:

Currently, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare professionals to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time.

#### Variant Evaluation:

Evaluation and categorization of variants are performed using published American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations as a guideline.<sup>(9)</sup> Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools are interpreted with caution and professional clinical judgement.

Rarely, incidental findings or secondary findings may implicate another predisposition or presence of active disease. Incidental findings may include, but are not limited to, results related to the sex chromosomes. These findings will be carefully reviewed to determine whether they will be reported.

#### Clinical Reference

1. George JN, Nester CM. Syndromes of thrombotic microangiopathy. *N Engl J Med*. 2014;371(7):1654-1666
2. Go RS, Winters JL, Leung N, et al. Thrombotic microangiopathy care pathway: A consensus statement for the Mayo Clinic Complement Alternative Pathway-Thrombotic Microangiopathy (CAP-TMA) Disease-Oriented Group. *Mayo Clin Proc*. 2016;91(9):1189-1211
3. Noris M, Bresin E, Mele C, Remuzzi G: Genetic atypical hemolytic-uremic syndrome. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews* [Internet]. University of Washington, Seattle; 2007. Updated September 23, 2021. Accessed December 2, 2025. Available at [www.ncbi.nlm.nih.gov/books/NBK1367/](http://www.ncbi.nlm.nih.gov/books/NBK1367/)

4. Magliulo EK, Ravipati P. C3 Glomerulopathy: A Current Perspective in an Evolving Landscape. *Glomerular Dis.* 2024;4(1):200-210. Published 2024 Oct 30. doi:10.1159/000542354
5. Kavanagh D, Goodship TH. Atypical hemolytic uremic syndrome, genetic basis, and clinical manifestations. *Hematology Am Soc Hematol Educ Program.* 2011;2011:15-20. doi:10.1182/asheducation-2011.1.15
6. Bernabeu-Herrero ME, Jimenez-Alcazar M, Anter J, et al. Complement factor H, FHR-3 and FHR-1 variants associate in an extended haplotype conferring increased risk of atypical hemolytic uremic syndrome. *Mol Immunol.* 2015;67(2 Pt B):276-286. doi:10.1016/j.molimm.2015.06.021
7. Nishimura J, Yamamoto M, Hayashi S, et al. Genetic variants in C5 and poor response to eculizumab. *N Engl J Med.* 2014;370(7):632-639. doi:10.1056/NEJMoa1311084
8. Durey MA, Sinha A, Togarsimalemath SK, Bagga A. Anti-complement-factor H-associated glomerulopathies. *Nat Rev Nephrol.* 2016;12(9):563-578. doi:10.1038/nrneph.2016.99
9. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015;17(5):405-424

## Performance

### Method Description

Capture-based and amplicon-based next-generation sequencing (NGS) are performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as other regions that have known disease-causing variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for deletions-insertions (delins) less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. NGS based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. See [Targeted Genes and Methodology Details for Atypical Hemolytic Uremic Syndrome / Thrombotic Microangiopathy / Complement 3 Glomerulopathy Gene Panel](#) for details regarding the targeted genes analyzed for each test and specific gene regions not routinely covered. (Unpublished Mayo method)

Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genes analyzed: *ADAMTS13*, *C3*, *C5* [Chr9(GRCh37):g.123759950-123759973 only], *CD46* (*MCP*), *CFB*, *CFH*, *CFHR1*, *CFHR2*, *CFHR3*, *CFHR4*, *CFHR5*, *CFI*, *DGKE*, *MMACHC*, *THBD*

### PDF Report

Supplemental

# Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome (aHUS)/Thrombotic Microangiopathy (TMA) /Complement 3 Glomerulopathy (C3G) Gene Panel, Varies

## Day(s) Performed

Varies

## Report Available

21 to 28 days

## Specimen Retention Time

Whole blood: 25 days (if available); Extracted DNA: 3 months

## Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

## Fees & Codes

### Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

### Test Classification

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

### CPT Code Information

81404

81479

81479 (if appropriate for government payers)

### LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AHUGP	aHUS/TMA/C3G Gene Panel	99967-2

Result ID	Test Result Name	Result LOINC® Value
618017	Test Description	62364-5
618018	Specimen	31208-2
618019	Source	31208-2
618020	Result Summary	50397-9
618021	Result	82939-0
618022	Interpretation	69047-9

## Test Definition: AHUGP

Atypical Hemolytic Uremic Syndrome  
(aHUS)/Thrombotic Microangiopathy (TMA)  
/Complement 3 Glomerulopathy (C3G) Gene  
Panel, Varies

618023	Additional Results	82939-0
618024	Resources	99622-3
618025	Additional Information	48767-8
618026	Method	85069-3
618027	Genes Analyzed	48018-6
618028	Disclaimer	62364-5
618029	Released By	18771-6