



# Test Definition: CGPH

Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies

## Overview

### Useful For

Customization of existing next-generation sequencing panels offered through Mayo Clinic Laboratories

Detection single nucleotide and copy number variants in a custom gene panel

Identification of a pathogenic variant may assist with diagnosis, prognosis, clinical management, familial screening, and genetic counseling for a hereditary condition

### Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
G145	Hereditary Custom Gene Panel Tier 1	No, (Bill Only)	No
G146	Hereditary Custom Gene Panel Tier 2	No, (Bill Only)	No
G147	Hereditary Custom Gene Panel Tier 3	No, (Bill Only)	No
G148	Hereditary Custom Gene Panel Tier 4	No, (Bill Only)	No
G149	Hereditary Custom Gene Panel Tier 5	No, (Bill Only)	No
G150	Hereditary Custom Gene Panel Tier 6	No, (Bill Only)	No

### Genetics Test Information

For more information, see Method Description and the following:

- [Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Epilepsy Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Hereditary Cancer Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Nephrology Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel](#)

### Testing Algorithm

Pricing for this test is based on the number of genes selected (1, 2-14, 15-49, 50-100, 101-500, and greater than 500)

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and their corresponding CPT codes. For more information see [Custom Gene Panel Ordering](#).

**Special Instructions**

- [Informed Consent for Genetic Testing](#)
- [Custom Gene Panel Ordering](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Molecular Genetics: Hereditary Custom Gene Panel Patient Information](#)
- [Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Hereditary Cancer Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Epilepsy Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Nephrology Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel](#)
- [Targeted Genes and Methodology Details for Inborn Errors of Immunity](#)

**Method Name**

Sequence Capture and Next-Generation Sequencing (NGS)/Polymerase Chain Reaction (PCR), Sanger Sequencing or Multiplex Ligation-Dependent Probe Amplification (MLPA)

**NY State Available**

Yes

**Specimen****Specimen Type**

Varies

**Ordering Guidance**

This test **requires** the creation of a unique Gene List ID that directs the laboratory to test the genes requested.

To create the **required Gene List ID** for your Custom Gene Panel, navigate to:

[-Custom Gene Ordering Tool](#)

[-Custom Gene Ordering Tutorial](#)

For answers to frequently asked questions, see [Custom gene ordering](#) on MayoClinicLabs.com.

Patients who have had a previous bone marrow transplant from an allogeneic donor should not have testing performed on blood, bone marrow, or saliva because any results generated will reflect the genome of the donor rather than the recipient. Testing on patients who have an active hematologic malignancy or hematologic disorder with clonal proliferation may identify both somatic mutations and germline variants, which may result in test failure or necessitate follow-up testing to determine whether the detected variant is germline or somatic. For these patients, testing a skin

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biopsy or cultured fibroblasts is recommended. For instructions for testing patients who have received a bone marrow transplant or have an active hematologic disorder, call 800-533-1710. For more information see Cautions.

Targeted testing for familial variants (also called site-specific or known mutation testing) is available under FMTT / Familial Variant, Targeted Testing, Varies. Call 800-533-1710 to obtain more information about this testing option.

**Shipping Instructions**

Specimen preferred to arrive within 96 hours of collection.

**Necessary Information**

[Molecular Genetics: Hereditary Custom Gene Panel Patient Information](#) is strongly recommended. Testing may proceed without the patient information; however, it aids in providing a more thorough interpretation. Ordering healthcare professionals are strongly encouraged to complete the form and send it with the specimen.

**Specimen Required**

**Specimen Type:** Whole blood

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

**Container/Tube:**

**Preferred:** Lavender top (EDTA) or yellow top (ACD)

**Acceptable:** Any anticoagulant

**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 4 days/Frozen 4 days

**Forms**

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy of the consent is on file.

The following documents are available:

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

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

2. [Molecular Genetics: Hereditary Custom Gene Panel Patient Information](#)

3. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:

-[Neurology Specialty Testing Client Test Request](#) (T732)

-[Renal Diagnostics Test Request](#) (T830)

-[Biochemical Genetics Test Request](#) (T798)

-[Cardiovascular Test Request](#) (T724)

**Specimen Minimum Volume**

See Specimen Required

**Reject Due To**

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

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**Specimen Stability Information**

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

**Clinical & Interpretive****Clinical Information**

This test can be used to customize genetic testing panels offered at Mayo Clinic Laboratories. Individual genes can be added or removed to an existing genetic testing panel. Additionally, this test can be used to create your own custom single gene or multi-gene panel or to combine existing panels within the same disease state.

**Note:** Any genes added to the custom panel must be from the same disease state. Only one Gene List ID may be submitted per Custom Gene Panel, Hereditary order. The Gene List ID can be created using the [Custom Gene Ordering tool](#) (see Ordering Guidance).

**Reference Values**

An interpretive report will be provided.

**Interpretation**

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(1) 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 amplified for sequencing or deletion and duplication analysis

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as a result of technical limitations of the assay, including regions of homology, high GC (guanine-cytosine) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

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

**Deletion/Duplication Analysis:**

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) may not be detected.

This test is not designed to detect low levels of mosaicism or to differentiate between somatic mutations 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.

For detailed information regarding gene specific performance and technical limitations, see the appropriate Targeted Gene and Methodology details in Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent 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-Policy:**

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.(1) 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 should be interpreted with caution and professional clinical judgment.

**Clinical Reference**

1. 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. doi:10.1038/gim.2015.30

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## Performance

### Method Description

Next-generation sequencing (NGS) and/or Sanger sequencing are performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as some other regions that have known disease-causing variants. The human genome reference GRCh37/hg19 build is 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 less than 40 base pairs (bp), above 95% for deletions up to 75 bp, and insertions up to 47 bp. NGS and/or a polymerase chain reaction-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 amplified for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC (guanine-cytosine) content, and repetitive sequences. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.(Unpublished Mayo method)

For details regarding the specific gene regions not routinely covered, see the appropriate information:

[-Targeted Genes and Methodology Details for Cardiovascular/Connective Tissue/Dyslipidemia/Cerebrovascular/Primary Ciliary Dyskinesia Custom Gene Panel](#)

[-Targeted Genes and Methodology Details for Epilepsy Custom Gene Panel](#)

[-Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel](#)

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[-Targeted Genes and Methodology Details for Neurologic Disorders Custom Gene Panel](#)

[-Targeted Genes and Methodology Details for the Nuclear Mitochondrial Disorders Custom Gene Panel](#)

### PDF Report

Supplemental

### Day(s) Performed

Varies

### Report Available

28 to 35 days

### Specimen Retention Time

Whole blood: 28 days (if available); Extracted DNA: 3 months

### Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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## 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

CPT codes are based on the gene content of the custom gene panel. Refer to the [Custom Gene Ordering Tool](#) for custom gene panel specific CPT code information.

81165 (if appropriate)

81166 (if appropriate)

81167 (if appropriate)

81162 (if appropriate)

81201 (if appropriate)

81216 (if appropriate)

81218 (if appropriate)

81223 (if appropriate)

81249 (if appropriate)

81252 (if appropriate)

81286 (if appropriate)

81292 (if appropriate)

81295 (if appropriate)

81298 (if appropriate)

81307 (if appropriate)

81317 (if appropriate)

81319 (if appropriate)

81321 (if appropriate)

81329 (if appropriate)

81351 (if appropriate)

81403 (if appropriate)

81404 (if appropriate)

81405 (if appropriate)

81406 (if appropriate)

81407 (if appropriate)

81408 (if appropriate)

81430 (if appropriate)

81431 (if appropriate)

81432 (if appropriate)

# Test Definition: CGPH

Custom Gene Panel, Hereditary,  
 Next-Generation Sequencing, Varies

- 81435 (if appropriate)
- 81437 (if appropriate)
- 81440 (if appropriate)
- 81441 (if appropriate)
- 81443 (if appropriate)
- 81448 (if appropriate)
- 81479 (if appropriate)
- 81189 (if appropriate)
- 81419 (if appropriate)

### LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CGPH	Custom Gene Panel, Hereditary	105259-6

Result ID	Test Result Name	Result LOINC® Value
MG135	Gene List ID	48018-6
610422	Test Description	62364-5
606046	Specimen	31208-2
606047	Source	31208-2
606040	Result Summary	50397-9
606041	Result	82939-0
606042	Interpretation	69047-9
610423	Resources	99622-3
606043	Additional Information	48767-8
606044	Method	85069-3
610424	Genes Analyzed	48018-6
606045	Disclaimer	62364-5
606048	Released By	18771-6
620157	Additional Results	82939-0