



1-800-533-1710

CGD and Phagocyte Disorder GenePanel

PHCGD

PATIENT NAME PHCGD, NORMAL					ORDER NUMBER P413000488
PATIENT ID X100498913	DATE OF BIRTH 10/30/1990	AGE 34 Y	SEX Male	REQUESTED BY PROVIDER UNKNOWN	
COLLECTED 5/13/2025, 6:00 AM	RECEIVED 5/14/2025, 12:17 PM	REPORTED 5/14/2025, 1:26 PM			
The collected, received, and reported dates and times on the report are in the time zone of the performing location.				CLIENT ORDER NUMBER X100498913	
7028846 MCL RochesterCampus Rochester MN 55901				CLIENT MRN C7028846-0006434	

TEST DESCRIPTION

Evaluation of 20 genes associated with phagocytic disorders and chronic granulomatous disease (CGD).

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Negative

RESULT

No reportable variants were detected.

INTERPRETATION

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Please note, this test is not designed to differentiate between somatic and germline variants. Thus, individuals with a clonal hematologic proliferation may have somatic variants detected by the methodology used for this testing. In addition, the presence of a clonal population comprising a high percentage of the peripheral white blood cell population may obscure germline findings, resulting in a false negative. Also, if an individual 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. If the individual tested here has a clonal hematologic disorder, submission of an alternate specimen type (e.g. skin biopsy or fibroblast culture) for repeat genetic testing may be useful to assess for germline variants. Clinical correlation is recommended.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

A genetic consultation may be of benefit.

FOLLOW-UP OPTION(S)

Reflex to Whole Exome Sequencing:

These results were interpreted from exome sequencing data analyzed for the specific gene(s) indicated. If clinically indicated, reflex analysis and interpretation of the whole exome using existing data can be completed by ordering WESPR/Whole Exome Sequencing Panel Reflex, Varies. Note: this reflex test cannot be ordered if the current testing was performed on a prenatal specimen.



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METHOD

Next generation sequencing (NGS) and/or Sanger sequencing was 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 pathogenic variants. NGS and/or a polymerase chain reaction (PCR)-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed. 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 indels less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. See the Genes Analyzed field for a list of genes tested. A supplemental droplet digital PCR method was used to test for the presence of the c.75_76del; p.Tyr26Hisfs*26 (delta GT) disease-causing variant in NCF1.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See www.mayocliniclabs.com (TEST ID PHCGD) for details regarding genes with regions not routinely covered.

GENES ANALYZED

CEBPE, CFTR, CSF2RA, CSF2RB, CTSC, CYBA, CYBB, CYBC1, FERMT3, G6PD, ITGB2, MRTFA, NCF1, NCF2, NCF4, RAC2, SLC35C1, SLC7A7, SMARCD2 and WDR1

DISCLAIMER

Clinical Correlations

The gene(s) analyzed by this test may have more than one associated phenotype or inheritance pattern. Additionally, the genetic variants detected may have reduced penetrance, variable expressivity, or different classifications based on disease state. Classification and reporting of the variants detected is based on the intended disease state of the test ordered. For more information regarding the phenotypic spectrum which may be involved for a specific gene, see OMIM (www.ncbi.nlm.nih.gov/omim) or GeneReviews (www.genereviews.org).

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.



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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(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

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, but assay performance below the minimum acceptable criteria or for failed regions will be noted. The clinical phenotype observed in this individual and/or family may be due to genetic variants not targeted by this test. 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 for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed 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. Please refer to the Targeted Genes and Methodology Details for the Phagocytic Disorders and Chronic Granulomatous Disease Gene Panel, Varies in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

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) may not be detected.

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



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See www.mayocliniclabs.com (TEST ID PHCGD) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation

Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

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 U.S. Food and Drug Administration.

RELEASED BY

Ann Marie Moyer, M.D., Ph.D.

IS THIS BONE MARROW

No

Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus

Lab Director : NIKOLA A BAUMANN

PHD

CLIA Certificate : 24D0404292

Address : 200 FIRST STREET SW

ROCHESTER MN 55905