

Overview

Useful For

Providing a comprehensive genetic evaluation for patients with a personal or family history suggestive of primary B-cell deficiencies and related disorders

Patients with B-cell immunodeficiency disorders who may have other clinical presentations, besides the humoral immune defect, such as inflammatory bowel disease, autoimmunity, or other as indicated above

Establishing a diagnosis of a B-cell deficiency or related disorder, in some cases, allowing for appropriate management and surveillance for disease features based on the gene involved

Identifying variants within genes known to be associated with increased risk for disease features allowing for predictive testing of at-risk family members

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_PMS2	PGL_PMS2C (Bill Only)	No	No
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Genetics Test Information

This test includes next-generation sequencing and supplemental Sanger sequencing to test for variants in the *AICDA*, *BLNK*, *BTK*, *CD79A*, *CD79B (B29)*, *CARD11*, *CD19*, *CD27*, *CD40*, *CD40LG*, *CD81*, *CR2 (CD21)*, *CXCR4*, *GATA2*, *ICOS*, *IGHM*, *IGLL1 (Lambda5)*, *IKZF1 (IKAROS)*, *LRBA*, *LRRC8A*, *MALT1*, *MS4A1 (CD20)*, *NFKB2*, *PIK3R1*, *PIK3CD*, *PLCG2*, *PRKCD*, *RNF168*, *SH2D1A*, *TCF3 (E47)*, *TNFRSF13B (TACI)*, *TNFRSF13C*, *TNFSF12 (TWEAK)*, and *UNG* genes.

Identification of a pathogenic variant may assist with prognosis, clinical management, familial screening, and genetic counseling.

Testing Algorithm

When clinical history of defective immunoglobulin class switching is provided, PMS2 gene analysis will be performed on whole blood or DNA submitted samples only via Sanger sequencing and multiplex ligation-dependent probe amplification at an additional charge.

Due to lower concentration of DNA yielded from alternate specimen sources, _PMS2 cannot be performed on any sample type other than whole blood or DNA extracted from whole blood.

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

Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Blood Spot Collection Card-Spanish Instructions](#)
- [Primary Immunodeficiencies Patient Information](#)
- [Blood Spot Collection Card-Chinese Instructions](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Blood Spot Collection Instructions](#)

Method Name

Custom Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Supplemental Sanger Sequencing

NY State Available

Yes

Specimen**Specimen Type**

Varies

Ordering Guidance

The genes on this panel are primarily associated with B cell defects. If suspecting or considering a combined immunodeficiency involving T cells and other cellular defects, or EBV-related primary immunodeficiencies, order SCDGP / Severe Combined Immunodeficiency Panel (63 genes), Next-Generation Sequencing, Varies. SCDGP includes the *RAG1*, *RAG2*, *IKBKG* genes among others that have a combined T and B cell immunodeficiency phenotype.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Mutation, Targeted Testing, Varies.

Necessary Information

1. [Primary Immunodeficiencies Patient Information](#) (T791) is strongly recommended, but not required, to be filled out and sent with the specimen. This information aids in providing a more thorough interpretation of test results. Ordering providers are strongly encouraged to complete the form and send it with the specimen.
2. Include physician name and phone number with specimen.

Specimen Required

Due to lower concentration of DNA yielded from alternate specimen sources, *_PMS2* cannot be performed on any specimen type other than whole blood or DNA extracted from whole blood.

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:**Preferred:****Specimen Type:** Whole blood**Container/Tube:** Lavender top (EDTA)**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**Specimen Type:** Blood spot**Supplies:** Card-Blood Spot Collection Filter Paper (T493)**Container/Tube:****Preferred:** Collection card (Whatman Protein Saver 903 Paper)**Acceptable:** Whatman FTA Classic paper, PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card**Specimen Volume:** 2 to 5 Blood spots**Collection Instructions:**

1. An alternative blood collection option for a patient 1 year of age or older is a fingerstick. See [Dried Blood Spot Collection Tutorial](#) for how to collect blood spots via fingerstick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated**Additional Information:**

1. Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.
2. For collection instructions, see [Blood Spot Collection Instructions](#).
3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777).
4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800).

Specimen Type: Peripheral blood mononuclear cells (PBMC)**Container/Tube:** Cell pellet**Collection Instructions:** Send as a suspension in freezing medium or cell pellet frozen on dry ice.**Specimen Stability Information:** Frozen**Specimen Type:** Cultured fibroblasts**Container/Tube:** T-75 or T-25 flask**Specimen Volume:** 1 Full T-75 or 2 full T-25 flasks

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

Additional Information: Indicate the tests to be performed on the fibroblast culture cells. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

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. Tubes of culture media can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Extracted DNA

Container/Tube: 2 mL screw top tube

Specimen Volume: 100 µL (microliters)

Collection Instructions:

1. The preferred volume is 100 µL at a concentration of 250 ng/µL
2. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred)/Ambient/Refrigerated

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 in Special Instructions:

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

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

2. [Primary Immunodeficiencies Patient Information](#) (T791) in Special Instructions

Specimen Minimum Volume

Whole blood: 1 mL

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

Primary B-cell disorders/humoral immunodeficiencies are characterized by an insufficient number of B cells or impaired functioning/differentiation of B cells. B-cell disorders account for approximately two-thirds of all genetic primary immunodeficiency disorders (PIDD) and may result in decrease or dysfunction of one or more isotypes of immunoglobulin, leading to increased susceptibility to infection, particularly bacterial infections such as sinopulmonary infections, gastrointestinal infections, otitis, skin infections, and conjunctivitis. In the absence of infection, patients may be asymptomatic and, thus, difficult to diagnose. In addition, primary B-cell disorders may result in lymphoproliferative disorders or be associated with autoimmune (AI) manifestations, including AI cytopenias, AI endocrine disorders, and AI enteropathy among others.

There are several PIDD that also have an associated T-cell and/or other cellular immunodeficiency, in addition to the B-cell defects.

In some disorders with agammaglobulinemia or hypogammaglobulinemia, patients may have reduced numbers of B cells, resulting in a severe reduction in all antibody isotypes. Often, they present in the first few years of life with recurrent bacterial infections, a severe life-threatening bacterial infection (ie, meningitis, sepsis), and decreased lymphoid tissue (ie, small adenoids, tonsils, and lymph nodes in X-linked agammaglobulinemia, due to Bruton tyrosine kinase [*BTK*] gene variants). Inheritance can be either X-linked (eg, due to variants in *BTK*), or autosomal recessive (eg, *IGHM*, *CD79A*, *CD79B*, *IGLL1*, *BLNK*, *LRRC8A*, and *PIK3R1*). B-cell lymphopenia with hypogammaglobulinemia can also be observed in WHIM syndrome (warts, hypogammaglobulinemia, infections, and myelokathexis), which results from pathogenic gain-of-function variants in the *CXCR4* gene. These patients also have severe peripheral neutropenia (absolute neutrophil count <500) with evidence of myelokathexis (neutrophil retention) in the bone marrow. In addition to recurrent infections (sinopulmonary, urinary tract, omphalitis, deep soft tissue abscesses, skin), patients are also susceptible to warts and condyloma acuminata due to human papillomavirus infection.

Common variable immunodeficiency (CVID) is the most common adult humoral immunodeficiency disorder with an incidence of approximately 1:25,000 to 1:50,000. CVID may present with frequent and unusual infections during early childhood, adolescence, or adulthood. As per current diagnostic criteria, CVID is not considered in children younger than 4 years of age. In addition, a significant proportion of patients may have autoimmune or inflammatory manifestations, enlarged lymphoid tissues, granulomas, and an increased susceptibility to cancer. These patients typically have normal numbers of B cells (<5% of CVID patients have less than 1% of B cells, which are considered to be due to early B-cell defects) but have impaired terminal differentiation, resulting in decreased levels of IgG and IgA, with or without a decrease in IgM. Over two-thirds of patients have quantitative defects in switched memory B cells. Some patients may also have quantitative and functional T-cell defects or NK-cell deficiency. Patients with decreased naive T-cell numbers are considered to have late-onset combined immunodeficiency (LOCID). Genetic variants have been identified in several genes, including *ICOS*, *TNFRSF13B* (TACI), *CD19*, *TNFRSF13C* (BAFFR), *MS4A1* (CD20), *CR2* (CD21), *CD81*, *LRBA*, *NFKB2*, *IKZF1* (IKAROS), among others, in a subset of CVID patients. However, the majority of these patients have unknown genetic defects and may have oligogenic or polygenic causes of disease.

Dysgammaglobulinemias including hyper-IgM syndrome and selective antibody deficiencies may also occur where a patient is either lacking a specific immunoglobulin isotype (eg, selective IgA deficiency) or a specific vaccine antibody

response (impaired pneumococcal polysaccharide responsiveness) or may have an elevated/normal IgM level. Selective deficiencies (ie, IgA deficiency, IgG deficiency) may be due to variants in genes encoding immunoglobulin heavy or light chains. Selective IgA deficiency (slgAD) is the most common PIDD with an incidence of 1:200 to 1:1000, depending on the cohort studied. Most patients with slgAD are asymptomatic though some may have frequent infections. There is also a higher incidence of celiac disease in this group. Most patients with selective antibody deficiencies are treated if they have frequent infections in addition to impaired vaccine antibody responses. Some patients with slgAD may have autoantibodies to IgA. Hyper IgM syndrome (mostly commonly due to variants in *CD40LG* but also due to other genes, eg, *CD40*, *AICDA*, *PI3KCD*, *UNG*) is characterized by an inability to switch from the production of IgM-type antibodies to IgG, IgA, or IgE isotypes. These individuals typically have a normal number of B cells. Patients with *CD40L* and *CD40* deficiency tend to present with severe opportunistic infections more reminiscent of a cellular immunodeficiency and, therefore, may also be considered as combined immunodeficiencies.

Primary B-cell disorders may also result in lymphoproliferative diseases characterized by dysgammaglobulinemia/hypogammaglobulinemia, persistent or severe complications of Epstein-Barr virus (including hemophagocytic lymphohistiocytosis), and lymphoproliferative disorders (including malignant lymphomas). Lymphomas that are associated with these disorders are typically high-grade B-cell lymphomas, non-Hodgkin type, extranodal, and often involve the intestine. Inflammatory bowel disease has also been associated with some forms. Inheritance of these lymphoproliferative diseases can be X-linked or autosomal recessive. For example, X-linked lymphoproliferative disease (XLP) is due to pathogenic variants in *SH2D1A* (XLP-1), while autosomal recessive lymphoproliferative syndrome 2 is caused by pathogenic variants in *TNFRSF7*, which encodes CD27. Some of these lymphoproliferative disorders clinically manifest following infection, especially with Epstein-Barr virus.

Post-meiotic segregation disorder, due to pathogenic variants in *PMS2*, leads to defective class switching from IgM and results in low serum IgG and IgA with elevated IgM. Patients also often demonstrate cafe-au-lait macules and are predisposed to several types of malignancy due to Lynch syndrome. *PMS2* testing will be performed only for patients who demonstrate defective class switching.

Table. Genes included in this panel

Gene symbol (alias)	Protein	OMIM	Incidence	Inheritance	Phenotype disorder
<i>AICDA</i>	Single-stranded DNA cytosine deaminase	605257	Unknown	AR	Immunodeficiency with hyper IgM, type 2
<i>BLNK</i>	B-cell linker protein isoform 1	604515	Unknown	AR	Agammaglobulinemia
<i>BTK</i>	Tyrosine-protein kinase BTK isoform 1	300300	1-9/million	XL	X-linked agammaglobulinemia
<i>CD79A</i>	B-cell antigen receptor complex-associated protein alpha chain isoform 1 precursor	112205	Unknown	AR	Agammaglobulinemia
<i>CD79B (B29)</i>	B-cell antigen receptor	147245	Unknown	AR	Agammaglobulinemia

	complex-associated protein beta chain isoform 1 precursor				
<i>CARD11</i>	Caspase recruitment domain-containing protein 11	607210		AR/AD	Immunodeficiency 11 (AR), B-cell expansion with NFKB and T-cell energy (AD)
<i>CD19</i>	B-lymphocyte antigen CD19 isoform 2 precursor	107265	Unknown	AR	Common variable immunodeficiency (CVID) 3
<i>CD27 (TNFRSF7)</i>	CD27 antigen precursor	186711		AR	Lymphoproliferative syndrome 2 (CD27 deficiency)
<i>CD40</i>	Tumor necrosis factor receptor superfamily member 5 isoform 1 precursor	109535	Unknown	AR	Immunodeficiency with hyper IgM
<i>CD40LG</i>	CD40 ligand	300386	2/million males	XL	Immunodeficiency with X-linked hyper IgM
<i>CD81</i>	CD81 antigen isoform 1	186845	Unknown	AR	Common variable immunodeficiency (CVID) 6
<i>CR2 (CD21)</i>	Complement receptor type 2 isoform 1 precursor	120650	Unknown	AR	Common variable immunodeficiency (CVID) 7
<i>CXCR4</i>	C-X-C chemokine receptor type 4 isoform b	162643		AD	Myelokathexis, isolated, WHIM syndrome (AD)
<i>GATA2</i>	Endothelial transcription factor GATA-2 isoform 1	137295		AD	Immunodeficiency 21, Emberger syndrome, susceptibility to acute myeloid Leukemia and myelodysplastic syndrome
<i>ICOS</i>	Inducible T-cell costimulator precursor	604558	Unknown	AR	Common variable immunodeficiency (CVID) 1
<i>IGHM</i>	IMMUNOGLOBULIN HEAVY CHAIN CONSTANT REGION MU	147020	Unknown	AR	Agammaglobulinemia 1
<i>IGLL1</i>	Immunoglobulin	146770	Unknown	AR	Agammaglobulinemia

<i>(LAMBDA-5)</i>	lambda-like polypeptide 1 isoform a precursor				
<i>IKZF1</i> <i>(IKAROS)</i>	DNA-binding protein Ikaros isoform 2	603023		AD with incomplete penetrance	Late-onset B-cell PID
<i>LRBA</i>	Lipopolysaccharide-responsive and beige-like anchor protein isoform 2	606453	Unknown	AR	Common variable immunodeficiency (CVID) 8 with autoimmunity
<i>LRRC8A</i>	Volume-regulated anion channel subunit LRRC8A	608360	Unknown	AD	Agammaglobulinemia
<i>MALT1</i>	Mucosa-associated lymphoid tissue lymphoma translocation protein 1 isoform a	604860		AR	Immunodeficiency 12
<i>MS4A1</i> <i>(CD20)</i>	B-lymphocyte antigen CD20	112210		AR	Common variable immunodeficiency (CVID) 5
<i>NFKB2</i>	Nuclear factor NF-kappa-B p100 subunit isoform a	164012	Unknown	AD	Common variable immunodeficiency (CVID) 10
<i>PIK3CD</i>	Phosphatidylinositol 4,5-bisphosphate 3-kinase catalytic subunit delta isoform	602839	Unknown	AD	Immunodeficiency 14, hyper IgM
<i>PIK3R1</i>	Phosphatidylinositol 3-kinase regulatory subunit alpha isoform 1	171833	Unknown	AR	Agammaglobulinemia
<i>PLCG2</i>	1-Phosphatidylinositol 4,5-bisphosphate phosphodiesterase gamma-2	600220	Rare	AD	Autoinflammation, antibody deficiency, and immune dysregulation syndrome familial cold autoinflammatory syndrome
<i>PRKCD</i>	Protein kinase C delta type	176977	Unknown	AR	Autoimmune lymphoproliferative syndrome, type III
<i>RNF168</i>	E3 ubiquitin-protein ligase RNF168	612688		AR	RIDDLE syndrome

<i>SH2D1A</i>	SH2 domain-containing protein 1A isoform 1	300490	1/million males	XL	X-linked lymphoproliferative syndrome
<i>TCF3 (E47)</i>	Transcription factor E2-alpha isoform E12	147141		AD	Agammaglobulinemia 8
<i>TNFRSF13B (TACI)</i>	Tumor necrosis factor receptor superfamily member 13B	604907	Unknown	AD or AR	Common variable immunodeficiency (CVID) 2, immunoglobulin A deficiency
<i>TNFRSF13C</i>	Tumor necrosis factor receptor superfamily member 13C	606269	Unknown	AD or AR	Common variable immunodeficiency (CVID) 4
<i>TNFSF12 (TWEAK)</i>	Tumor necrosis factor ligand superfamily member 12 proprotein	602695		AD	Low IgM and IgA
<i>UNG</i>	Uracil-DNA glycosylase isoform UNG2	191525	Unknown	AR	Immunodeficiency with hyper IgM syndrome, type 5

AD=autosomal dominant
 AR=autosomal recessive
 XL=X-linked

Reference Values

An interpretive report will be provided.

Interpretation

Evaluation and categorization of variants is performed using the most recent published American College of Medical Genetics and Genomics (ACMG) recommendations as a guideline.(1) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

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

Cautions

Clinical Correlations:

Some individuals who have involvement of one or more of the genes on the panel may have a variant that is not identified by the methods performed (eg, promoter variants, deep intronic variants). The absence of a variant, therefore, does not eliminate the possibility of primary B-cell deficiency or a related disorder. 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.

For predictive testing of asymptomatic individuals, it is often useful to first test an affected family member. Identification of a pathogenic variant in an affected individual allows for more informative testing of at-risk individuals.

Technical Limitations:

Next-generation sequencing may not detect all types of genetic variants. The variant detection software has lower detection efficiency for insertion/deletion variants as compared to single nucleotide variants. Therefore, small deletions and insertions greater than 8 nucleotides in length may not be detected by this test. Copy number variations are not currently reported for any of the genes on this panel. Additionally, rare alterations (ie, polymorphisms) may be present that could lead to false-negative or false-positive results. In some cases, DNA variants of undetermined significance may be identified. If results do not match clinical findings, consider alternative methods for analyzing these genes, such as Sanger sequencing or large deletion/duplication analysis.

If the patient has had an allogeneic blood or bone marrow transplant or a recent (ie, <6 weeks from time of sample collection) heterologous blood transfusion, results may be inaccurate due to the presence of donor DNA. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants-Policy:

At this time, it is not standard practice for the laboratory to systematically review likely pathogenic variants or variants of uncertain significance that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time. Consultation with a healthcare provider, or team of healthcare providers, with expertise in genetics and primary immunodeficiencies, is recommended for interpretation of this result.

A list of benign and likely benign variants detected is available from the lab upon request.

Contact the laboratory if additional information is required regarding the transcript or human genome assembly used for the analysis of results.

Clinical Reference

- [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 May;17\(5\):405-424](#)
- Picard C, Gaspar HB, Al-Herz W, Bousfina A, et al: International Union of Immunological Societies: 2017 Primary Immunodeficiency Disease Committee Report on Inborn Errors of Immunity, J Clin Immunol. 2018;38:96-128
- Conley ME, Dobbs AK, Farmer DM, et al: Primary B cell immunodeficiencies: Comparisons and contrasts. Ann Rev Immunol. 2009;27:199-277
- Park MA, Li JT, Hagan JB, et al: Common Variable Immunodeficiency: a new look at an old disease. Lancet. 2008;372:489-502
- Ameratunga R, Brewerton M, Slade C, et al: Comparison of diagnostic criteria for common variable immunodeficiency disorder. Front Immunol. 2014;415(5):1-9
- Schaffer AA, Salzer U, Hammarstrom L, et al: Deconstructing common variable immunodeficiency by genetic analysis. Curr Opin. Genetics and Dev 2007;7:201-212

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7. Conley ME: Early defects in B cell development. *Curr Opin Allergy Clin Immunol* .2002;2:517-522
 8. Notarangelo LD, Lanzi G, Peron S, et al: Defects of class-switch recombination. *J Allergy Clin Immunol* .2006;117:855-864
 9. Bousfiha AA, Jeddane L, Ailal F, et al: A phenotypic approach for IUIS PID classification and diagnosis: guidelines for clinicians at the bedside. *J Clin Immunol*. 2013;33:1078-1087

Performance

Method Description

Next-generation sequencing (NGS) is performed using an Illumina instrument with paired-end reads. The DNA is prepared for NGS using a custom Agilent SureSelect Target Enrichment System. Data is analyzed with a bioinformatics software pipeline. Supplemental Sanger sequencing may be performed occasionally in regions where NGS is insufficient for data capture or not specific enough to correctly identify a variant.(Unpublished Mayo method)

Genes analyzed: *AICDA*, *BLNK*, *BTK*, *CD79A*, *CD79B (B29)*, *CARD11*, *CD19*, *CD27*, *CD40*, *CD40LG*, *CD81*, *CR2 (CD21)*, *CXCR4*, *GATA2*, *ICOS*, *IGHM*, *IGLL1 (Lambda5)*, *IKZF1 (IKAROS)*, *LRBA*, *LRRC8A*, *MALT1*, *MS4A1 (CD20)*, *NFKB2*, *PIK3R1*, *PIK3CD*, *PLCG2*, *PRKCD*, *RNF168*, *SH2D1A*, *TCF3 (E47)*, *TNFRSF13B (TACI)*, *TNFRSF13C*, *TNFSF12 (TWEAK)*, and *UNG*.

PMS2: Bidirectional sequence analysis is performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the *PMS2* gene. Additionally, gene dosage analysis (multiplex ligation-dependent probe amplification) is used to test for the presence of large deletions and duplications in this gene.(Vaughn CP, Hart J, Samowitz WS, Swensen JJ: Avoidance of pseudogene interference in the detection of 3' deletions in *PMS2*. *Hum Mutat*. 2011;32:1063-1071)

PDF Report

No

Day(s) Performed

Monday

Report Available

28 to 56 days

Specimen Retention Time

Extracted DNA: 2 months

Performing Laboratory Location

Rochester

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. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81443

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
BCLGP	B-cell Deficiency PID Gene Panel	97565-6

Result ID	Test Result Name	Result LOINC® Value
BA3886	Gene(s) Evaluated	48018-6
BA3887	Result Summary	50397-9
BA3888	Result Details	82939-0
BA3889	Interpretation	69047-9
BA3890	Additional Information	48767-8
BA3891	Method	85069-3
BA3892	Disclaimer	62364-5
BA3893	Reviewed by	18771-6