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MAYO CLINIC LABORATORIES

Test Definition: BCELL

B-Cell and Antibody Deficiency Gene Panel, Varies

Overview

Useful For

Providing a comprehensive genetic evaluation for patients with a personal or family history suggestive of an inherited primary B-cell disorder or humoral immunodeficiency

Establishing a diagnosis of a primary B-cell disorder or humoral immunodeficiency, allowing for appropriate management and surveillance for disease features based on the gene or variant involved

Identifying variants within genes known to be associated with primary B-cell disorders or humoral immunodeficiencies, allowing for predictive testing of at-risk family members

Reflex Tests

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

Genetics Test Information

This test utilizes next-generation sequencing to detect single nucleotide and copy number variants in 61 genes associated with inherited B-cell disorders and humoral immunodeficiency: *ADA, ADA2, AICDA, ATP6AP1, BLNK, BTK, CARD11, CD19, CD27, CD40, CD40LG, CD70, CD79A, CD79B, CD81, CDCA7, CTLA4, CR2, CXCR4, DCLRE1C, DNMT3B, GATA2, ICOS, IGHM, IGLL1, IKBKG, IKZF1, IKZF3, IL21, IL21R, IRF2BP2, KDM6A, KMT2A, KMT2D, LIG1, LRBA, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, RAG1, RAG2, RNF168, SEC61A1, SH2D1A, SH3KBP1, SLC39A7, TCF3, TNFRSF13B, TNFRSF13C, TNFSF12, TNFSF13, TOP2B, TRNT1, UNG,* and *XIAP.* See Targeted Genes and Methodology Details for B-Cell and Antibody Deficiency Gene Panel for details regarding the targeted gene regions evaluated by this test.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, recurrence risk assessment, familial screening, and genetic counseling for inherited B-cell disorders and humoral immunodeficiency.

Testing Algorithm

Skin biopsy:

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
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Informed Consent for Genetic Testing (Spanish)
- Blood Spot Collection Instructions
- Combined Immunodeficiency, Severe Combined Immunodeficiency, and B-Cell/Antibody Deficiency Patient Information
- Targeted Genes and Methodology Details for B-Cell and Antibody Deficiency Gene Panel

Method Name

Sequence Capture and Amplicon-Based Next-Generation Sequencing (NGS), Polymerase Chain Reaction (PCR), and Dosage Analysis by Droplet Digital Polymerase Chain Reaction (ddPCR)/Quantitative Real-Time Polymerase Chain Reaction (qPCR) and Sanger Sequencing as needed

NY State Available

Yes

Specimen

Specimen Type

Varies

Ordering Guidance

Patients who have had a previous bone marrow transplant from an allogenic 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 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.

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 Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis disease state for step 1 on the Custom Gene Ordering Tool.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Additional Testing Requirements

For cord blood specimens: Maternal cell contamination (MCC) studies are available. **Order MATCC /** Maternal Cell Contamination, Molecular Analysis, Varies **on both the cord blood and maternal specimens under separate order numbers**. Cord blood testing will proceed without MCC studies, but results may be compromised if MCC is present.

Specimen Required

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

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:
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: Cultured fibroblasts
Source: Skin
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy. Cultured cells from a prenatal specimen will not be accepted.
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 mcL at a concentration of 75 ng/mcL.
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.

Specimen Type: Blood spot
Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: PerkinElmer 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 older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample.
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. Blood spot specimens are acceptable but not recommended. Multiple extractions will be required to obtain sufficient yield for supplemental analysis, and there is significant risk for test failure due to insufficient DNA.
2. Due to lower concentration of DNA yielded from blood spot, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.
3. For collection instructions, see Blood Spot Collection Instructions
4. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
5. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

Specimen Type: Saliva
Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies:
DNA Saliva Kit High Yield (T1007)
Saliva Swab Collection Kit (T786)
Container/Tube:
Preferred: High-yield DNA saliva kit
Acceptable: Saliva swab
Specimen Volume: 1 Tube if using T1007 or 2 swabs if using T786
Collection Instructions: Collect and send specimen per kit instructions.
Specimen Stability Information: Ambient (preferred) 30 days/Refrigerated 30 days
Additional Information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

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)
-Informed Consent for Genetic Testing (Spanish) (T826)
2. Combined Immunodeficiency, Severe Combined Immunodeficiency, and B-Cell/Antibody Deficiency Patient Information

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

Primary B-cell disorders and humoral immunodeficiencies are characterized by an insufficient number of B cells or the impaired functioning or differentiation of B cells. B-cell disorders account for approximately half to two-thirds of all genetic primary immunodeficiency disorders (PIDD). They may result in a 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.

Primary immunodeficiency disorders that are primarily antibody deficiencies fall into four main categories:
1. Agammaglobulinemias, which are characterized by severe reduction in all serum immunoglobulin isotypes with profoundly decreased or absent B cells
2. Common variable immunodeficiency (CVID)-like diseases that are characterized by severe reduction in at least two serum immunoglobulin isotypes with normal or low number of B cells
3. Hyper-IgM syndromes, which are characterized by severe reduction in serum IgG and IgA with normal or elevated IgM and normal numbers of B cells
4. A mixed group of isotype, light chain, or functional antibody deficiencies generally with normal numbers of B cells
In addition, there are several PIDD that also have an associated T-cell or other cellular immunodeficiency as well as B-cell defects.

Agammaglobulinemia typically presents 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*), autosomal dominant (eg, *TCF3*, *TOP2B*), or autosomal recessive (eg, *IGHM*, *CD79A*, *CD79B*, *IGLL1*, *BLNK*, and *PIK3R1*).

Common variable immunodeficiency (CVID) is the most common adult humoral immunodeficiency disorder with an incidence of approximately 1:10,000 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. 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 <1% B cells, which is 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 natural killer (NK) cell deficiency. Patients with decreased naive T-cell numbers are considered to have late-onset combined immunodeficiency. Genetic variants have been identified in several genes, including *ICOS*, *TNFRSF13B* (*TACI*), *CD19*, *TNFRSF13C* (*BAFFR*), *MS4A1* (*CD20*), *CR2* (*CD21*), *CD81*, *LRBA*, *NFKB2*, and *IKZF1* (*IKAROS*) in a subset of CVID patients. However, most of these patients have unknown genetic defects and may have oligogenic or polygenic causes of disease.

Hyper IgM syndrome is characterized by an inability to switch from the production of IgM-type antibodies to IgG, IgA, or IgE isotypes. The condition is most often caused by variants in *CD40LG*, but variants in other genes (eg, *CD40*, *AICDA*, *PI3KCD*, *UNG*) have also been reported to cause disease. 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.

Selective antibody deficiencies may occur when a patient is either lacking a specific immunoglobulin isotype (eg, selective IgA deficiency or IgG deficiency) or a specific vaccine antibody response (impaired pneumococcal polysaccharide responsiveness). Selective deficiencies may be due to variants in genes encoding immunoglobulin heavy or light chains. Selective IgA deficiency (sIgAD) is the most common PIDD with an incidence of 1:200 to 1:1000, depending on the cohort studied. Most patients with sIgAD 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 sIgAD may have autoantibodies to IgA.

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 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.

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.

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 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.

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-leukoreduced 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. Due to broadening genetic knowledge, it is possible that the laboratory may discover new information of relevance to the patient. Should that occur, the laboratory may issue an amended report.

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.

Rarely, incidental or secondary findings may implicate another predisposition or presence of active disease. These findings will be carefully reviewed to determine whether they will be reported.

Clinical Reference

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11. Bousfiha A, Moundir A, Tangye SG, et al. The 2022 update of IUIS phenotypical classification for human inborn errors of immunity. J Clin Immunol. 2022;42(7):1508-1520. doi:10.1007/s10875-022-01352-z

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 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 and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. A supplemental PCR-based method is used to detect a large deletion in *IKBKG*, and a supplemental droplet digital PCR method is used to detect deletions and duplications in *IGHM*. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

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 B-Cell and Antibody Deficiency Gene Panel for details regarding the targeted gene regions identified by this test.(Unpublished Mayo method)

Genes analyzed: *ADA, ADA2, AICDA, ATP6AP1, BLNK, BTK, CARD11, CD19, CD27, CD40, CD40LG, CD70, CD79A, CD79B, CD81, CDCA7, CTLA4, CR2, CXCR4, DCLRE1C, DNMT3B, GATA2, ICOS, IGHM, IGLL1, IKBKG, IKZF1, IKZF3, IL21, IL21R, IRF2BP2, KDM6A, KMT2A, KMT2D, LIG1, LRBA, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, RAG1, RAG2, RNF168, SEC61A1, SH2D1A, SH3KBP1, SLC39A7, TCF3, TNFRSF13B, TNFRSF13C, TNFSF12, TNFSF13, TOP2B, TRNT1, UNG,* and *XIAP.*

PDF Report
Supplemental

Day(s) Performed
Varies

Report Available
28 to 42 days

Specimen Retention Time
Whole blood: 28 days (if available); Saliva: 30 days (if available); Extracted DNA: 3 months; Blood spots: 1 year (if available)

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

81443
88233-Tissue culture, skin, solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
BCELL	Bcell/Antibody Deficiency GenePanel	97565-6

Result ID	Test Result Name	Result LOINC® Value
620107	Test Description	62364-5
620108	Specimen	31208-2
620109	Source	31208-2
620110	Result Summary	50397-9
620111	Result	82939-0
620112	Interpretation	69047-9
620113	Additional Results	82939-0
620114	Resources	99622-3
620115	Additional Information	48767-8
620116	Method	85069-3
620117	Genes Analyzed	82939-0
620118	Disclaimer	62364-5
620119	Released By	18771-6