Alpha-1-antitrypsin (A1A) is a protein that inhibits the enzyme neutrophil elastase. It is predominantly synthesized in the liver and secreted into the bloodstream. The inhibition function is especially important in the lungs to protect against excess tissue degradation. Tissue degradation due to A1A deficiency is associated with an increased risk for early onset panlobar emphysema, which initially affects the lung bases (as opposed to smoking-related emphysema, which presents with upper lung field emphysema). Patients may become symptomatic in their 30s and 40s. The most frequent symptoms reported in a NIH study of 1,129 patients with severe deficiency (mean age 46 years) included cough (42%), wheezing (65%), and dyspnea with exertion (84%). Many patients were misdiagnosed as having asthma. It is estimated that approximately 1/6 of all lung transplant patients are for A1A deficiency. Liver disease can also occur, particularly in children; it occurs much less commonly than emphysema in adults.

A1A deficiency is a relatively common disorder in Northern European Caucasians. The diagnosis of A1A deficiency is initially made by quantitation of protein levels in serum followed by determining specific allelic variants by isoelectric focusing (IEF). While there are many different alleles in this gene, only 3 are common. The 3 major alleles include: M (full functioning, normal allele), S (associated with reduced levels of protein) and Z (disease causing mutation associated with liver disease and premature emphysema), with S and Z accounting for the majority of the abnormal alleles detected in affected patients. As a codominant disorder, both alleles are expressed. An individual of SZ or Snull genotype may have a small increased risk for emphysema (but not liver disease) because of slightly reduced protein levels. On the other hand, an individual with the ZZ genotype is at greater risk for early onset liver disease and premature emphysema. Smoking appears to hasten development of emphysema by 10-15 years. These individuals should be monitored closely for lung and liver function.

Historically, IEF has been the primary method for characterizing variants. However, in some cases the interpretation of IEF is difficult and prone to error. DNA-based assays are now available for the characterization of deficiency alleles in patients with A1A deficiency. See “Alpha-1-Antitrypsin Testing Algorithm” Figure 1, page 6, August 2002 Communiqué.

Useful For

• Confirmation of clinical diagnosis of A1A deficiency and determination of the specific allelic variant.
  Genotyping also provides some insight as to the possible course of disease.
• Prenatal diagnosis in at-risk pregnancies

Interpretation

The quantitative level of A1A should correlate with the genotype and a level will be determined for each patient. If the genotyping and quantitative level are discordant, IEF will be done to clarify the results. The report will include specimen information, pedigree (when appropriate), assay information, background information, and estimate of carrier risk based on test results. The report will also include quantitative levels of A1A. A1A phenotype will be included when necessary.

Normal individuals will have “non-Z, non-S” genotypes.
See “Alpha-1-Antitrypsin Outcomes Chart” Figure 2, page 7, August 2002 Communiqué.
Cautions

- Direct DNA testing will not detect all known variants within the A1A gene. A negative test result for the S and Z allele, therefore, does not rule out the diagnosis of A1A deficiency. The laboratory will identify discordant genotype and quantitative levels of A1A and perform phenotyping to clarify the interpretation.
- Test results should be interpreted in the context of A1A quantitation, clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information provided to us is inaccurate or incomplete.
- Because the Z allele is relatively frequent in the Caucasian population (about 1 in 50), genetic counseling is recommended for individuals and families who have a diagnosis of A1A deficiency.
- Medical genetic consultation is available for all cases and is particularly indicated in complex cases or in situations in which the diagnosis is atypical or uncertain.

References


Method

A PCR-based assay is used to detect the Z and S allele within the A1A gene. Other A1A variants will not be detected by this assay. A1A serum levels are measured by immunonephelometry. The serum level will be reported with the genotyping result. Any genotyping results that are discordant with the serum level will be phenotyped by isoelectric focusing. (Pierce JA: Hereditary pulmonary emphysema. In Emery and Rimoin’s Principles and Practices of Medical Genetics, Vol II, 3rd edition. Edited by DL Rimoin, JM Connor, RE Pyeritz. New York, Churchill Livingstone, 1997, pp 2727-2750; Behring Nephelometer II Operations Instruction Manual. Dade Behring, Inc, Newark, DE)

Specimen Required: Both blood and serum are required for this test.

Specimens should include a “Molecular Genetics Congenital Disorders Request Form” (Supply T245) or a “Mayo Connect Additional Test Information” form (Supply T357) with information including relevant clinical and family history information. Specimens must arrive within 96 hours of draw.

Blood

Draw blood in a lavender-top (EDTA) tube(s) or a yellow-top (ACD) tube(s), and send 3.0 mL of whole blood in the original VACUTAINER(S). Invert several times to mix blood. Forward unprocessed whole blood promptly at ambient temperature only.

NOTE: Label specimen appropriately (blood).

Serum

Draw blood in a plain, red-top tube(s) or an SST tube(s). Spin down and send 1.0 mL of serum refrigerated.

NOTE: Label specimen appropriately (serum).

Reference Values: ALPHA-1-ANTITRYPsin GENOTYPE
An interpretive report will be issued.

ALPHA-1-ANTITRYPsin
100-190 mg/dL

Analytic Time: 5 days
Days Set Up: Monday through Friday
Fee: $248.10
CPT Code: 83890/Molecular isolation and extraction
83898x2/PCR amplification
83912/Interpretation and report
Antineutrophil cytoplasmic antibodies (ANCA) occur in patients with autoimmune vasculitis including Wegener's granulomatosis (WG), microscopic polyangiitis (MPA), or organ-limited variants thereof such as pauci-immune necrotizing glomerulonephritis.

ANCA react with enzymes in the cytoplasmic granules of human neutrophils including PR3, myeloperoxidase (MPO), elastase, and cathepsin G.

Autoantibodies to PR3 occur in patients with WG (both classical WG and WG with limited end-organ involvement) and produce a characteristic pattern of granular cytoplasmic fluorescence on ethanol-fixed neutrophils called the cANCA pattern.

Antibodies to MPO occur predominately in patients with MPA and produce a pattern of perinuclear cytoplasmic fluorescence on ethanol-fixed neutrophils called the pANCA pattern.

Autoantibodies to PR3 and MPO also can be detected by enzyme immunoassay (EIA) methods and are referred to as PR3 ANCA and MPO ANCA, respectively.

Evaluating patients suspected of having autoimmune vasculitis, both WG and MPA

Positive results for PR3 ANCA and cANCA or pANCA are consistent with the diagnosis of WG, either systemic WG with respiratory and renal involvement or limited WG with more restricted end-organ involvement.

Positive results for MPO ANCA and pANCA are consistent with the diagnosis of autoimmune vasculitis including MPA or pauci-immune necrotizing glomerulonephritis.

A positive result for PR3 ANCA or MPO ANCA has been shown to detect 89% of patients with active WG or MPA (with or without renal involvement) with fewer than 1% false-positive results in patients with other diseases.

The ANCA Vasculitis Panel cannot be relied upon exclusively to establish the diagnosis of autoimmune vasculitis (WG or MPA):
- Some patients with WG or MPA may not have a measurable titer of antibodies detected by this panel of tests.
- Some of these patients have antibodies to other neutrophil antigens not included in the panel, eg, neutrophil elastase.
Test Title: Antineutrophil Cytoplasmic Antibodies Vasculitis Panel, Serum #83012

References

Method
PR3 ANCA and MPO ANCA are measured by commercial microtiter enzyme immunoassays (Scimedx Corp., Denville, NJ. 07834;2). Results are expressed in arbitrary units of EU/mL by comparison with a multipoint standard curve. These assays are approved for in vitro diagnostic use by FDA. cANCA and pANCA are measured by an in-house indirect immunofluorescence assay on ethanol-fixed human neutrophils.

Specimen Required: Draw blood in a plain, red-top tube(s). Spin down and send 0.5 mL of serum frozen in a plastic vial.

Reference Values:

MYELOPEROXIDASE ANTIBODIES
\[ \leq 5.0 \text{ EU/mL: Negative} \]
\[ \geq 5.1-400.0 \text{ EU/mL: Positive} \]

AUTOANTIBODIES TO PROTEINASE 3
\[ \leq 5 \text{ EU/mL: Negative} \]
\[ >5 \text{ EU/mL: Positive} \]

CYTOPLASMIC NEUTROPHIL ANTIBODIES
Negative
If positive for cANCA, results are titered.

Analytic Time: 2 days
Days Set Up: Monday through Saturday
Fee: $191.50
CPT Code: 83516/PR3
83520/MPO
86255/screen (if appropriate)
86256/titer (if appropriate)
Autoantibodies to Proteinase 3, Serum

#82965

Clinical

• PR3 antigen is a 29kD serine protease that exists as a protein triplet in human neutrophils.
• Patients with Wegener’s granulomatosis (WG) develop autoantibodies to the proteinase 3 (PR3) antigen of myeloid lysosomes.1

Useful For

• Evaluating patients suspected of having WG
• Since it is often not possible to distinguish clinically between WG and other forms of vasculitis, the test for PR3 ANCA should be employed with other serologic tests in the initial diagnostic evaluation of such patients (See #83012 “Anti-neutrophil Cytoplasmic Antibodies Vasculitis Panel, Serum”).

Interpretation

• A positive test for PR3 ANCA has a high positive predictive value for WG; fewer than 2% of positive results occur in patients who do not have WG.2
• The presence of autoantibodies to PR3, called PR3 antineutrophil cytoplasmic antibodies (ANCA), is a specific diagnostic indicator of WG and is found in more than 80% of patients with active WG.2
• A negative result for PR3 ANCA diminishes the likelihood that a patient has active WG, but approximately 20% of patients with WG may test negative for PR3 ANCA.

Cautions

• While the presence of PR3 ANCA is highly specific for WG, it is recommended that positive test results obtained by enzyme immunoassay be confirmed by another testing method.3 This is best accomplished by testing for cANCA and pANCA by indirect immunofluorescence microscopy (See #9441 “Cytoplasmic Neutrophil Antibodies, Serum”). Simultaneous presence of PR3 ANCA and cANCA has a specificity >99% for WG.3
• Testing for PR3 ANCA is not recommended for monitoring disease activity and response to treatment.

References


Method

PR3 antibodies are measured using a commercial enzyme immunoassay kit. Polystyrene microtiter plate wells coated with purified PR3 antigen are used to capture antibody from the patient sera. Alkaline phosphatase conjugated antihuman IgG is used as a detection protein. PR3 antibodies are measured by quantitation of the color development generated when disodium p-nitrophenyl phosphate substrate is reacted with the alkaline phosphatase conjugate. (Anti-Proteinase Semi-quantitative Kit: PR396. Scimedx Corporation. 1/13/00)

Specimen Required: Draw blood in a plain, red-top tube(s). Spin down and send 0.5 mL of serum frozen in a plastic vial.

Reference Values: ≤5 EU/mL: Negative
>5 EU/mL: Positive

Analytic Time: 1 day
Days Set Up: Monday through Saturday
Fee: $111.30
CPT Code: 83516
B-type natriuretic peptide (brain natriuretic peptide [BNP]) is a 32 amino acid ringed peptide secreted by the heart to regulate blood pressure and fluid balance.\(^1\) BNP is stored in and secreted predominantly from membrane granules in the heart ventricles and is continuously released from the heart in response to both ventricle volume expansion and pressure overload.\(^2\)

The natriuretic peptide system and the renin-angiotensin system counteract each other in arterial pressure regulation. When arterial pressure decreases, the kidneys release renin, a small protein enzyme that circulates throughout the bloodstream. Angiotensinogen, a polypeptide released from the liver, is cleaved in the circulation by renin to form angiotensin I. This biologically inactive decapeptide is cleaved in turn by a second enzyme (angiotensin converting enzyme) to form active angiotensin II. Angiotensin II is a vasoconstrictor that increases the peripheral resistance of the arterioles, thus increasing arterial pressure.

Both BNP and ANP (atrial natriuretic peptide) are activated by atrial and ventricular distension due to increased intracardiac pressure. These peptides have both natriuretic and diuretic properties: they raise sodium and water excretion by increasing the glomerular filtration rate and inhibiting sodium reabsorption by the kidney. The natriuretic peptides counteract the effects of renin secretion, causing a reduction of blood pressure and extracellular fluid volume.\(^3\)

The New York Heart Association (NYHA) developed a functional classification system for congestive heart failure (CHF) consisting of 4 stages based on the severity of the symptoms. Various studies have demonstrated that circulating BNP concentrations increase with the severity of CHF based on the NYHA classification.\(^4\)\(^5\)\(^6\)

- **Aids in the diagnosis of CHF**
- **The role of BNP in monitoring CHF therapy is under investigation.**

**Interpretation**

<table>
<thead>
<tr>
<th>Functional Class</th>
<th>5th – 95(^{th}) Percentile</th>
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<tr>
<td>I</td>
<td>15 to 499 pg/mL</td>
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<tr>
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<tr>
<td>III</td>
<td>38 to &gt;1300 pg/mL</td>
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<tr>
<td>IV</td>
<td>147 to &gt;1300 pg/mL</td>
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<tr>
<td>All CHF</td>
<td>22 to &gt;1300 pg/mL</td>
<td>360 pg/mL</td>
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Elevation in BNP can occur due to right heart failure with cor pulmonale (200-500 pg/mL), pulmonary hypertension (300-500 pg/mL) and acute pulmonary embolism (150-500 pg/mL). Elevations also occur in patients with acute coronary syndromes.

**Cautions**

Lack of elevations have been reported if CHF is very acute (first hour) or with ventricular inflow obstruction (hypertrophic obstructive cardiomyopathy, mitral stenosis, atrial myxoma).
Test Title: B-Type Natriuretic Peptide (BNP), Plasma

#80022

References

Method
BNP is measured using a fluorescence immunoassay system manufactured by Biosite. The assay implemented in the Central Clinical Laboratory uses plasma specimens in which EDTA is the anticoagulant. After addition of a plasma specimen to the specimen port of the test device a predetermined quantity of plasma moves by capillary action into a reaction chamber and is allowed to react with fluorescent antibody conjugates within the reaction chamber to form a reaction mixture. After an incubation period, the reaction mixture flows through the device detection lane. Complexes of the analyte and fluorescent antibody conjugates are captured on discrete zones in the detection lane. Excess plasma specimen washes the unbound fluorescent antibody conjugates from the detection lane into a waste reservoir. The concentration of the analyte in the specimen is proportional to the fluorescence bound to the detection lane. (Package insert: Triage BNP Test Product Insert. Biosite Inc., San Diego, 92121, Rev C., December 2001)

Specimen Required: Draw blood in a lavender-top (EDTA) tube(s). Spin down, remove plasma from cells within 4 hours of draw and send 1.0 mL of EDTA plasma frozen in plastic vial.

NOTE: Include patient's age and sex on request form.

Reference Values:

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<td>≤93</td>
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</table>

Analytic Time: 1 day
Days Set Up: Monday through Sunday
Fee: $150.00
CPT Code: 83520
The clinical symptoms of fatty acid beta-oxidation (FAO) disorders are similar and include hypoketotic, hypoglycemia, a Reye-like syndrome, myopathy (involving skeletal and/or heart muscle), and sudden death. Patients with any of these disorders are at risk of developing fatal metabolic decompensations following the acquisition of even common viral infections or prolonged fasting. Once diagnosed, these disorders can be treated by frequent meals, special diets, and cofactor/vitamin supplementation. The combined incidence of FAO disorders is approximately 1:13,000 live births.

Commonly used metabolite screens such as urine organic acids, plasma acylcarnitines, and plasma fatty acids are influenced by dietary factors and the clinical status of the patient, which can make the biochemical diagnosis of these disorders difficult. Fasting tests to provoke metabolic abnormalities in the patient pose a significant risk to the patient's health and must be performed under close medical observation in a hospital setting. Furthermore, even specimens collected during acute illness or a fasting test may only reveal nonspecific metabolite profiles. The FAO probe assay simulates a fasting test of the patient and offers an unequivocal evaluation of the mitochondrial FAO pathway under controlled conditions and at no risk to the patient. By assessing the complete FAO pathway, this assay is also more efficient than enzyme testing, which is limited to 1 catalytic activity per assay, and molecular genetic methods for rapid detection of common mutations, which are not completely informative for compound heterozygotes with an uncommon private mutation.

In vitro confirmation of biochemical diagnoses of the following FAO disorders:
• Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
• Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
• Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
• Trifunctional protein deficiency
• Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
• Carnitine palmitoyl transferase deficiency type II (CPT-II)
• Carnitine-acylcarnitine translocase (CACT) deficiency
• Multiple acyl-CoA dehydrogenase deficiency (glutaric acidemia type II)

In addition, the following organic acid disorders can be confirmed by this assay:
• 2-methylbutyryl-CoA dehydrogenase (SBCAD) deficiency
• Isobutyryl-CoA dehydrogenase (IBD) deficiency

Work is in progress to evaluate the applicability of this assay to the remaining disorders of fatty acid transport and mitochondrial oxidation.

Quantitative results of individual acylcarnitines are not diagnostic: interpretation is based on pattern recognition.

The report includes quantitative results along with an interpretation. The interpretation of abnormal acylcarnitine profiles includes an overview of the results and of their significance, a correlation to available clinical information, possible differential diagnoses, recommendations for additional biochemical testing and confirmatory studies if indicated, name and phone number of contacts who may provide these studies at Mayo Clinic or elsewhere, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.
Test Title: Fatty Acid Oxidation Probe Assay, Fibroblast Culture #81927

Cautions
Sometimes an abnormal acylcarnitine profile cannot differentiate between 2 disorders. In such instances, independent biochemical (eg, specific enzyme assay) or molecular genetic analyses are required.

References

Method
Skin fibroblasts are incubated with cell medium enriched with palmitic acid (C16:0 fatty acid) and L-carnitine. Cell lines deficient of 1 of the enzymes involved in fatty acid oxidation fail to metabolize acyl-CoA species, which accumulate in the cell medium as acylcarnitines. The medium is separated from the cells following the incubation. The cell pellet is used for protein determination and the medium is spotted and dried on filter paper. An acylcarnitine analysis is performed by tandem mass spectrometry (MS/MS) using a 1/4” filter paper punch, following the addition of isotopically labeled acylcarnitines as internal standards, extraction and derivatization to methyl esters. The assay is performed in triplicate (Matern D, Huey JC, Gregersen N, et al: In vitro diagnosis of short-chain acyl-CoA dehydrogenase (SCAD) deficiency. Pediatr Res 2001;49[Suppl 2]:185A).

Specimen Required:
Skin Biopsy
Skin biopsy (4-mm punch). Collect sterile biopsy in Eagle's minimum essential medium with penicillin, streptomycin, and nystatin (Supply T115). (SPECIMEN RECEIVED IN FORMALIN OR FIXATIVE PRESERVATIVE IS NOT ACCEPTABLE). Maintain sterility and forward promptly at ambient temperature only. See test #8482 “Fibroblast Culture.”

Cultured Fibroblasts
2 T25 flasks filled to neck with culture media. (SPECIMEN RECEIVED IN FORMALIN OR FIXATIVE PRESERVATIVE IS NOT ACCEPTABLE.) Maintain sterility and forward promptly at ambient temperature only.

Reference Values:

<table>
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<tr>
<th></th>
<th>C2 0.29 - 7.46 µmol/g protein</th>
<th>C12 &lt;0.40 µmol/g protein</th>
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<tbody>
<tr>
<td>C3</td>
<td>&lt;1.15 µmol/g protein</td>
<td>C12-OH ≤0.03 µmol/g protein</td>
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<td>C4</td>
<td>&lt;1.60 µmol/g protein</td>
<td>C14:1 &lt;0.11 µmol/g protein</td>
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<td>C5</td>
<td>&lt;1.02 µmol/g protein</td>
<td>C14 ≤0.21 µmol/g protein</td>
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<tr>
<td>C6</td>
<td>&lt;0.69 µmol/g protein</td>
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<td>≤0.05 µmol/g protein</td>
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<td>&lt;0.78 µmol/g protein</td>
<td>C16-OH ≤0.03 µmol/g protein</td>
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Analytic Time: 10 to 31 days
Days Set Up: Varies
Fee: $490.00 (When Fibroblast Culture is performed it is charged separately.)
CPT Code: 80500/Clinical pathology consultation
82017/Acylcarnitines; quantitative, each specimen
Specimen Source Identification
#82971

Clinical
For various reasons, the patient origin for a particular specimen may be questioned. This is especially true for paraffin-embedded material: labeling accuracy may be questioned or tissue from other sources may be included by mistake. Confirmation of the patient origin may be critical to the clinical work-up of that patient.

Molecular methods are now available to extract DNA from various sources, including paraffin-embedded material, and to compare the molecular fingerprint (genotype) of one specimen source with another one. Matching genotypes on multiple specimens suggest that they are derived from the same patient, whereas differences in genotype suggest different patient sources.

Useful For
Determining specimen origin when the patient identity of a specimen is in question

Interpretation
The report will include specimen information, assay information, and interpretation of test results.

Cautions
• This test is not intended for use in forensic cases; it should be applied to only those cases that have clinical implications.
• The laboratory is not structured for chain-of-custody documentation.

References

Method
A panel of microsatellite markers that recognize highly variable regions of human DNA is used in a polymerase chain reaction-based assay to compare the genotype for the specimen in question with that determined from DNA isolated from patient specimens of known identity. The markers tested include the dinucleotide repeats D17S250, D5S346, ACTC, D18S55, and D8S262. (Sano K, Takayanagi K, Kaneko T, et al: Application of short tandem repeat of genomic DNA and mitochondrial DNA for identification of mixed-up tissue specimens. Pathol Int 2000;50:1-6)

Specimen Required:
Specimen should include a “Molecular Genetics Congenital Disorders Request Form” (Supply T245) or a “MayoConnect Additional Test Information Form” (Supply T357) with relevant clinical and family history information. Specimen must arrive within 96 hours of draw.

**BLOOD**
Draw blood in a lavender-top (EDTA) tube(s) or a yellow-top (ACD) tube(s), and send 5.0 mL of EDTA or ACD whole blood in the original VACUTAINER(S). Invert several times to mix blood. Forward unprocessed whole blood promptly at ambient temperature only.

**TISSUE**
Send paraffin-embedded whole tissue block OR 4x10 micron thick sections plus 1 slide stained with hematoxylin-and-eosin.
Test Title: Specimen Source Identification
#82971

Analytic Time: 14 days
Days Set Up:
Paraffin-embedded tissue already mounted on slides received by Thursday will be set up on Monday.
To allow time for slide preparation if a paraffin block is sent, blocks must be received by
Wednesday for Monday setup
Fee:
$254.40
CPT Code:
83890 x 1/Molecular isolation or extraction
83894 x 10/Separation by gel electrophoresis
83898 x 10/PCR amplification
83912 x 1/Interpretation and report
Monoclonal antibodies are critical tools for detecting cellular antigens in various hematologic diseases and are used to provide critical diagnostic information. Monoclonal antibodies are also used as therapeutic agents in a variety of hematologic diseases. For example, anti-CD20 antibodies have been used to treat patients with B-cell malignant lymphomas and multiple myeloma. Other examples include:
- Anti-CD52 (Campath-1H): B-cell chronic lymphocytic leukemia and T-cell disorders
- Anti-CD22: hairy cell leukemia
- Anti-CD3 and T-cell malignancies
- Anti-CD33: acute myeloid leukemia

This list will undoubtedly expand over time to include other antibodies.

It may be necessary to document expression of these markers by the malignant cells prior to initiating the respective monoclonal antibody therapy. Expression of these markers may also be required for follow-up to monitor the impact of treatment on residual normal counterparts (eg, CD20-positive lymphocytes in patients treated with anti-CD20).

### Useful For
- Detecting cell-surface antigens on malignant cells that are potential therapeutic antibody targets
- Determining the eligibility of patients for monoclonal antibody therapies
- Monitoring response to the therapeutic antibody

### Interpretation
The immunophenotyping report will summarize the pattern of antigenic expression on malignant cells and, if necessary, the normal cellular counterpart(s) that correspond to the therapeutic monoclonal antibody target.

### Cautions
- The requesting physician must provide the following information:
  - therapeutic monoclonal antibody being used/considered
  - pertinent hematologic disease(s) that has/have been diagnosed/considered
  - any pertinent protocol requirements
- A complete diagnostic B-cell, T-cell, or acute immunophenotyping panel will not be performed and must be ordered separately (#3287 “Leukemia/Lymphoma Immunphenotyping by Flow Cytometry”). In some cases, a limited morphologic evaluation will be performed.

### References
Test Title: Therapeutic Antibody by Flow Cytometry #82977

Method
Flow cytometric immunophenotyping of peripheral blood, bone marrow, or tissue-derived lymphocytes is performed to assess the expression of the cell-surface antigen corresponding to the monoclonal antibody therapeutic target. Limited antibody panels will be used and may include:
- Anti-CD20 assessment - CD19/CD20/kappa/lambda or CD20/CD38/CD45 kappa/lambda
- Anti-CD52 assessment - CD19/CD3/CD52/kappa/lambda
- Anti-CD22 assessment - CD19/CD22/kappa/lambda
- Anti-CD33 assessment - CD13/CD33/CD45/CD34

Dependent on the therapeutic monoclonal antibody, appropriate panels will be developed.


Specimen Required: SUBMIT ONLY 1 OF THE FOLLOWING SPECIMENS:

Peripheral Blood
Draw blood in a yellow-top (ACD [solution B]) tube(s), and send 7.0 mL of ACD (solution B) peripheral blood. Do not transfer blood to other containers. Forward promptly at ambient temperature only.
SPECIMEN CANNOT BE FROZEN.
NOTE: 1. COLLECTION DATE IS REQUIRED ON REQUEST FORM FOR PROCESSING.
2. A PATHOLOGY/DIAGNOSTIC REPORT, THE NAME AND TELEPHONE NUMBER OF THE ORDERING PHYSICIAN, AND A BRIEF HISTORY ARE ESSENTIAL TO ACHIEVE A CONSULTATION FULLY RELEVANT TO THE ORDERING PHYSICIAN'S NEEDS.
3. Indicate blood on request form.
4. Label specimen appropriately (blood).
5. Please complete a "Hematopathology/Molecular Oncology Request Form" (Supply T241) or a "MayoConnect Additional Test Information Form" (Supply T357) and forward it with the specimen.

Bone Marrow
1.0-5.0 mL of ACD (solution B) bone marrow. Submission of bilateral specimens is not required. Include 5 air-dried fresh bone marrow smears, if possible. Forward promptly at ambient temperature only.
SPECIMEN CANNOT BE FROZEN.
NOTE: 1. COLLECTION DATE IS REQUIRED ON REQUEST FORM FOR PROCESSING.
2. A PATHOLOGY/DIAGNOSTIC REPORT, THE NAME AND TELEPHONE NUMBER OF THE ORDERING PHYSICIAN, AND A BRIEF HISTORY ARE ESSENTIAL TO ACHIEVE A CONSULTATION FULLY RELEVANT TO THE ORDERING PHYSICIAN'S NEEDS.
3. Indicate bone marrow on request form.
4. Label specimen appropriately (bone marrow).
5. Please complete a "Hematopathology/Molecular Oncology Request Form" (Supply T241) or a "MayoConnect Additional Test Information Form" (Supply T357) and forward it with the specimen.

Reference Values: Normal individuals have B-lymphocytes, T-lymphocytes, or myeloid cells that express the corresponding cell-surface antigen(s) in question.

Analytic Time: 1 day
Days Set Up: Monday through Sunday
Fee: $180.50
CPT Code: 85060/Blood smear, interp by physician
88180 x 4/Flow cytometry, each cell