Test Definition: LAD1
Leukocyte Adhesion Def. Type 1, B

Overview

Useful For
Aids in the diagnosis of leukocyte adhesion deficiency syndrome type 1, primarily in patients younger than 18 years of age
CD11a, CD11b, and CD18 phenotyping

Method Name
FlowCytometricImmunophenotyping
IncludesevaluationofmarkersCD11a/CD18andCD11b/CD18.

NY State Available
Yes

Specimen

Specimen Type
Whole Blood EDTA

Shipping Instructions
Specimens are required to be received in the laboratory weekdays and by 4 p.m. on Friday. Draw and package specimen as close to shipping time as possible.

It is recommended that specimens arrive within 24 hours of draw.

Samples arriving on the weekend and observed holidays may be canceled.

Necessary Information
Date and time of draw and physician name and phone number are required.

Specimen Required
For serial monitoring, we recommend that specimen draws be performed at the same time of day.

Container/Tube: Lavender top (EDTA)

Specimen Volume: 5 mL

Collection Instructions: Send specimen in original tube. Do not aliquot.

Specimen Minimum Volume
2 mL

Reject Due To

<table>
<thead>
<tr>
<th>Condition</th>
<th>Action</th>
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<tbody>
<tr>
<td>Gross hemolysis</td>
<td>Reject</td>
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<tr>
<td>Gross lipemia</td>
<td>Reject</td>
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</table>
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Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Whole Blood EDTA</td>
<td>Ambient</td>
<td>72 hours</td>
<td>PURPLE OR PINK TOP/EDTA</td>
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Clinical and Interpretive

Clinical Information

Leukocyte adhesion deficiency syndrome type 1 (LAD-1) is an autosomal recessive disorder caused by mutations in the common chain (CD18) of the beta2-integrin family. LAD-1 is clinically characterized by recurrent infections, impaired wound healing, delayed umbilical cord separation, persistent leukocytosis, and recurrent soft tissue and oral infections.

Each of the beta2-integrins is a heterodimer composed of an alpha chain (CD11a, CD11b, or CD11c) noncovalently linked to a common beta2-subunit (CD18). The alpha-beta heterodimers of the beta2-integrin family include LFA-1 (CD11a/CD18), Mac-1/CR3 (CD11b/CD18), and p150/95 (CD11c/CD18).(1-4) The CD18 gene, ITGB2, and its product are required for normal expression of the alpha-beta heterodimers. Therefore, defects in CD18 expression lead to either very low or no surface membrane expression of CD11a, CD11b, and CD11c.

Severe and moderate forms of LAD-1 exist, differing in the degrees of protein deficiency, which are caused by different ITGB2 mutations. Two relatively distinct clinical phenotypes of LAD-1 have been described. Patients with the severe phenotype (<1% of normal expression of CD18 on neutrophils) characteristically have delayed umbilical stump separation (>30 days), infection of the umbilical stump (omphalitis), persistent leukocytosis (>15,000/microliter) in the absence of overt active infection, and severe destructive gingivitis with periodontitis and associated tooth loss, and alveolar bone resorption. Patients with the moderate phenotype of LAD-1 (1%-30% of normal expression of CD18 on neutrophils) tend to be diagnosed later in life. Normal umbilical separation, lower risk of life-threatening infections, and longer life expectancy are common in these patients. However, leukocytosis, periodontal disease, and delayed wound healing are still very significant clinical features.

Patients with LAD-1 (and other primary immunodeficiency diseases) are unlikely to remain undiagnosed in adulthood. Consequently, this test should not be typically ordered in adults for LAD-1. However, it may be also used to assess immune competence by determining CD18, 11a, and 11b expression.

Reference Values

Normal (reported as normal or absent expression for each marker)

Interpretation

The report will include a summary interpretation of the presence or reduction in the level of expression of the individual markers (CD11a, CD11b, and CD18). Expression of the individual markers provides indirect information on the presence or absence of the CD11a/CD18 and CD11b/CD18 complexes.

Specimens obtained from patients with leukocyte adhesion deficiency syndrome type 1 (LAD-1) show significant reduction (moderate phenotype) or near absence (severe phenotype) of CD18 and its associated molecules, CD11a and CD11b, on neutrophils and other leukocytes.

CD11c expression also is low in LAD-1. The analytical sensitivity of the CD11c assay is insufficient to allow
interpretation of CD11c surface expression. Therefore, we test only for expression of CD18, CD11a, and CD11b.

**Cautions**

This test is typically not indicated in adults. For questions about appropriate test selection, call 800-533-1710.

Patients with normal beta2-integrin expression without functional activity have been described. Therefore, expression of CD18 alone is insufficient to exclude the diagnosis of leukocyte adhesion deficiency syndrome type 1 (LAD-1); functional assays (e.g., neutrophil chemotaxis, random migration assays) must be performed if the clinical suspicion is high.

**Clinical Reference**


**Performance**

**Method Description**

Flow cytometric immunophenotyping of peripheral blood is performed to evaluate the presence or absence of the CD11/CD18 complex using monoclonal antibodies directed against the CD11 isoforms, CD11a and CD11b, and CD18 antigens. (O’Gorman MR, McNally AC, Anderson DC, et al: A rapid whole blood lysis technique for the diagnosis of moderate or severe leukocyte adhesion deficiency [LAD]. Ann NY Acad Sci 1993;677:427-430)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Monday through Friday

**Do not send specimen after Thursday.** Specimen must be received by 10 a.m. on Friday.

**Analytic Time**

3 days

**Maximum Laboratory Time**

4 days
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Specimen Retention Time
4 days

Performing Laboratory Location
Rochester

Fees and 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 Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed using an analyte specific reagent. Its performance characteristics were 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.

CPT Code Information
86356 x 3

LOINC® Information

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<td>Leukocyte Adhesion Def. Type 1, B</td>
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