Overview

Useful For
Aiding in the diagnosis of lipopolysaccharide-responsive beige-like anchor protein (LRBA) deficiency

This test is not useful for identifying a carrier status for LRBA deficiency.

Genetics Test Information
The human lipopolysaccharide-responsive beige-like anchor protein (LRBA) gene is on chromosome 4.

Assessment of 109 patients with LRBA deficiency has shown 93 homozygous and 16 compound heterozygous alterations in the gene.

Alterations in the LRBA gene have been observed throughout the length of the gene and include the following main categories: Nonsense; missense; insertions, deletions, indels, and splice site alterations.

Highlights
The test determines the percentage and intensity of expression of lipopolysaccharide-responsive beige-like anchor (LRBA) protein on T cells and B cells in peripheral blood.

It can be used as a screening step prior to genetic testing for LRBA; to confirm the finding of an established disease-causing alteration in LRBA at the protein level; and to examine the effect of reported genetic variants of undetermined significance on LRBA protein expression.

It can help distinguish LRBA deficiency from conditions with overlapping clinical manifestations, including immune dysregulation and autoimmunity, such as immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX)-like syndromes; early onset hypogammaglobulinemia; common variable immune deficiency; inflammatory bowel disease; and autoimmune lymphoproliferative syndrome.

Method Name
Flow Cytometry

NY State Available
Yes

Specimen

Specimen Type
Whole Blood EDTA
Test Definition: LRBA
Lipopolysaccharide-Responsive Beige-Like Anchor Protein (LRBA) Deficiency, Blood

Ordering Guidance
This flow cytometry test is complementary to genetic testing.

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

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

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

Necessary Information
Ordering physician name and phone number are required.

Specimen Required
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send whole blood specimen in original tube. Do not aliquot.

Specimen Minimum Volume
1 mL

Reject Due To

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<td>Reject</td>
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<tr>
<td>Gross icterus</td>
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Specimen Stability Information

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<td>Ambient (preferred)</td>
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Clinical & Interpretive

Clinical Information
Lipopolysaccharide-responsive beige-like anchor protein (LRBA) deficiency is a rare autosomal recessive primary immunodeficiency disease (also known as inborn errors of immunity) caused by homozygous or compound heterozygous loss-of-function variants in the LRBA gene. It has a wide spectrum of clinical manifestations, including immune dysregulation and autoimmunity, inflammatory bowel disease, early-onset hypogammaglobulinemia, recurrent infections and organomegaly.

Reference Values
The appropriate reference values will be provided on the report.
Interpretation
The results are reported as the percentage and MFI (mean fluorescence intensity) of lipopolysaccharide-responsive beige-like anchor protein (LRBA) expression in T cells and B cells.

The majority of genetically confirmed cases of LRBA deficiency led to the absence of LRBA expression. Therefore, the lack of LRBA expression in T and B cells is consistent with LRBA deficiency. In this case, genetic analysis of LRBA to confirm the diagnosis and to identify the underlying variant will be recommended.

In addition, there are reported cases of LRBA deficiency where the protein is expressed but at lower intensity. Therefore, the expression of LRBA at diminished intensity could be due to a disease-causing LRBA variant, which would have to be confirmed or ruled out by genetic and functional analysis.

Cautions
No significant cautionary statements

Clinical Reference

Performance
Method Description
The lipopolysaccharide-responsive beige-like anchor (LRBA) protein expression assay is performed on EDTA whole blood. Samples are fixed, permeabilized and stained with antibodies specific for CD45, CD14, CD19, CD3, and CD56 along with either the LRBA antibody (unconjugated) or isotype control (unconjugated). A secondary reporter antibody is added to allow the assessment of LRBA and isotype control expression. Samples are then analyzed on a flow cytometer. LRBA expression is evaluated on the following populations: T-cells: (CD45+CD14negCD3+) and B-cells: (CD45+CD14negCD3negCD19+).(Unpublished Mayo method)

PDF Report
No
Test Definition: LRBA
Lipopolysaccharide-Responsive Beige-Like Anchor Protein (LRBA) Deficiency, Blood

Day(s) Performed
Monday through Friday

Report Available
2 to 4 days

Specimen Retention Time
4 days

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 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 US Food and Drug Administration.

CPT Code Information
86356 x 2

LOINC® Information

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