MAYO CLINIC LABORATORIES

# Test Definition: ALPRT

ALPORT Immunofluorescence

## Overview

### Useful For

Assisting in the diagnosis of hereditary nephritis (Alport syndrome)

### Special Instructions

- Renal Biopsy Patient Information
- Renal Biopsy Procedure for Handling Tissue for Light Microscopy (LM), Immunofluorescent Histology (IH), and Electron Microscopy (EM)

### Method Name

Direct Immunofluorescence

### NY State Available

Yes

## Specimen

### Specimen Type

Special

### Shipping Instructions

1. Advise shipping specimens in Styrofoam transportation coolers to avoid extreme hot or cold temperatures to ensure specimens are received at required specimen stability temperature.

2. Attach the green pathology address label included in the kit to the outside of the transport container.

### Necessary Information

**A pathology/diagnostic report is required.**

### Specimen Required

**Specimen Type:** Kidney tissue

**Supplies:** Renal Biopsy Kit (T231)

**Container/Tube:** Transport medium (Michel's or Zeus media), frozen tissue

**Specimen Volume:** Entire specimen

**Collection Instructions:**

1. For kidney cases, collect specimens according to the instructions in Renal Biopsy Procedure for Handling Tissue for Light Microscopy, Immunohistology, and Electron Microscopy

2. If standard immunoglobulin and complement immunofluorescence has already been performed, submit the residual frozen tissue (must contain glomeruli) on dry ice.

**Acceptable:** 2 Frozen tissue unstained positively charged glass slides (25- x 75- x 1-mm) per test ordered; sections 4-microns thick, submitted on dry ice.

**Specimen Type:** Skin tissue

**Container/Tube:** Transport medium (Michel's or Zeus media)

**Specimen Volume:** Entire specimen

**Collection Instructions:** Submit punch biopsy in Zeus/Michel's media.

### Forms

1. Renal Biopsy Patient Information Sheet

2. If not ordering electronically, complete, print, and send a Renal Diagnostics Test Request (T830) with the specimen.

### Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability

### Specimen Stability Information

| Specimen Type | Temperature | Time | Special Container |
|---|---|---|---|
| Special | Ambient (preferred) | | |
| | Frozen | | |
| | Refrigerated | | |

## Clinical & Interpretive

### Clinical Information

Alport syndrome is a hereditary disease of basement membrane collagen type IV. Variants in collagen IV alpha genes cause characteristic abnormal immunofluorescence staining patterns within the glomerular basement membrane. Alport syndrome is characterized by hematuria, proteinuria, progressive renal failure, and high-tone sensorineural hearing loss.

### Reference Values

An interpretive report will be provided.

### Interpretation

This test, (when not accompanied by a pathology consultation request) will be reported as: 1) normal pattern, 2) consistent with X-linked hereditary nephritis, or 3) consistent with autosomal hereditary nephritis.

If additional interpretation or analysis is needed, request PATHC / Pathology Consultation along with this test and send the corresponding renal pathology light microscopy and immunofluorescence (IF) slides (or IF images on a CD), electron microscopy images (prints or CD), and the pathology report.

### Cautions

Approximately one-third of patients with established hereditary nephritis based on typical ultrastructural findings and family history show loss of glomerular basement membrane or epidermal basement membrane staining for the alpha 5 chain of type IV collagen. Therefore, a normal staining pattern does not exclude the diagnosis of hereditary nephritis.

Because alpha 3 and alpha 4 chains of type IV collagen are not expressed in the epidermal basement membranes, patients with autosomal hereditary nephritis have preserved staining for alpha 5 on epidermal basement membranes and, therefore, skin biopsy cannot exclude autosomal hereditary nephritis.

### Clinical Reference

1. Kagawa M, Kishiro Y, Naito I, et al: Epitope-defined monoclonal antibodies against type-IV collagen for diagnosis of Alport's syndrome. Nephrol Dial Transplant. 1997 Jun;12(6):1238-1241

2. Hashimura Y, Nozu K, Nakanishi K, et al: Milder clinical aspects of X-linked Alport syndrome in men positive for the collagen IV alpha 5 chain. Kidney Int. 2014;85(5):1208-1213

3. Kamiyoshi N, Nozu K, Fu XJ, et al: Genetic, clinical, and pathologic backgrounds of patients with autosomal dominant Alport syndrome. Clin J Am Soc Nephrol. 2016 Aug 8;11(8):1441-1449

4. Said SM, Fidler ME, Valeri AM, et al: Negative staining for COL4A5 correlates with worse prognosis and more severe ultrastructural alterations in males with Alport syndrome. Kidney Int Rep. 2017;2(1):44-52

## Performance

### Method Description

Direct immunofluorescence staining on sections of fresh/frozen tissue.(Unpublished Mayo method)

### PDF Report
No

### Performing Laboratory Location
Rochester

## Fees & Codes

### 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
88346-primary IF

### LOINC® Information

| Test ID | Test Order Name | Order LOINC Value |
|---|---|---|
| ALPRT | ALPORT Immunofluorescence | In Process |

| Result ID | Reporting Name | LOINC® |
|---|---|---|
| 71285 | Interpretation | 50595-8 |
| 71268 | Participated in the Interpretation | No LOINC Needed |
| 71272 | Material Received | 81178-6 |
| 71271 | Gross Description | 22634-0 |
| 71269 | Report electronically signed by | 19139-5 |
| 71270 | Addendum | 35265-8 |
| 71619 | Disclaimer | 62364-5 |
| 71848 | Case Number | 80398-1 |